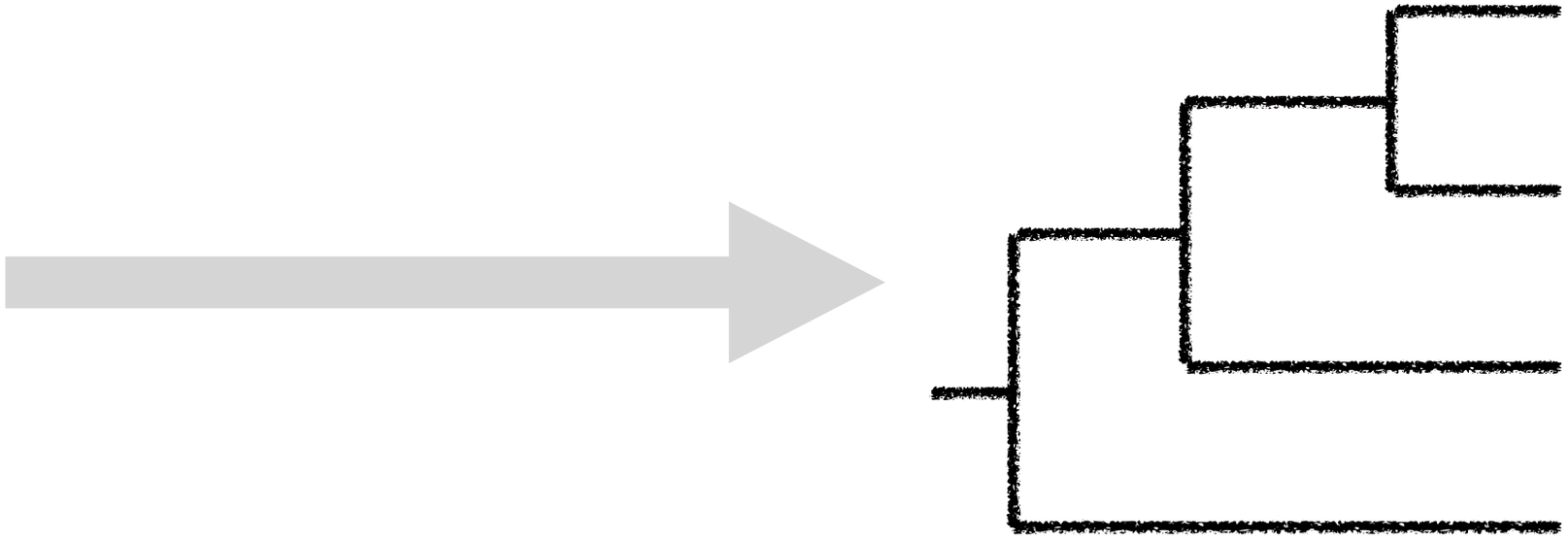
Lecture 14

Coalescent-based methods
Botany 563 — Spring 2022

- **Previous class check-up:**
    - We studied Bayesian phylogenetic inference
    - We practiced on MrBayes
- **Learning Objectives:** At the end of today's session, you will be able to
    - Explain the coalescent model on a species tree
    - Explain the steps in coalescent-based methods and the comparison with concatenation approaches
- **Pre-class work**
    - Read HAL 3.1 and 3.3
- **Important: start thinking about final project (due May 9th)!**

Phylogenetic inference

AAGTCTAG
AAGTCTAG
AACTCTAG
AATTCTAG



Phylogenetic inference

AAGTCTAG
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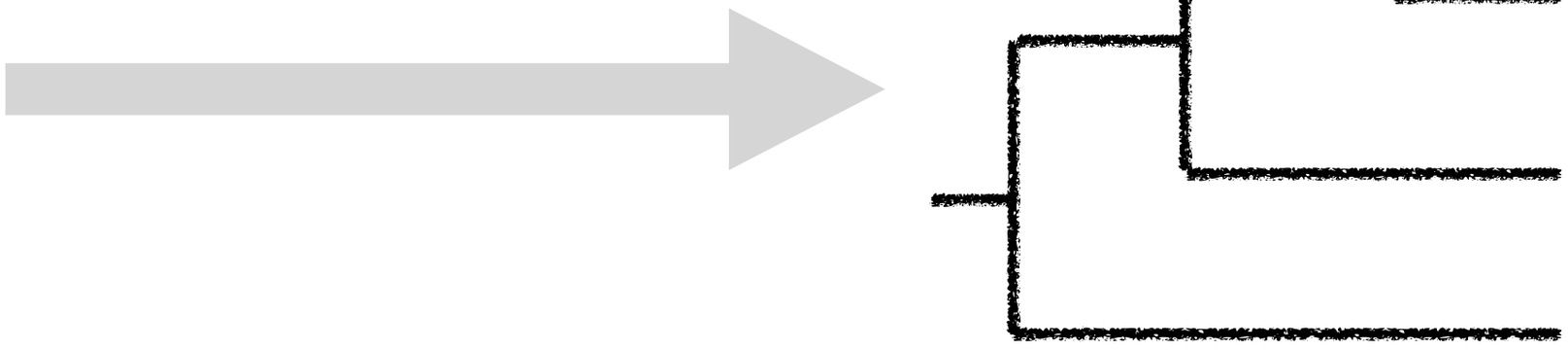
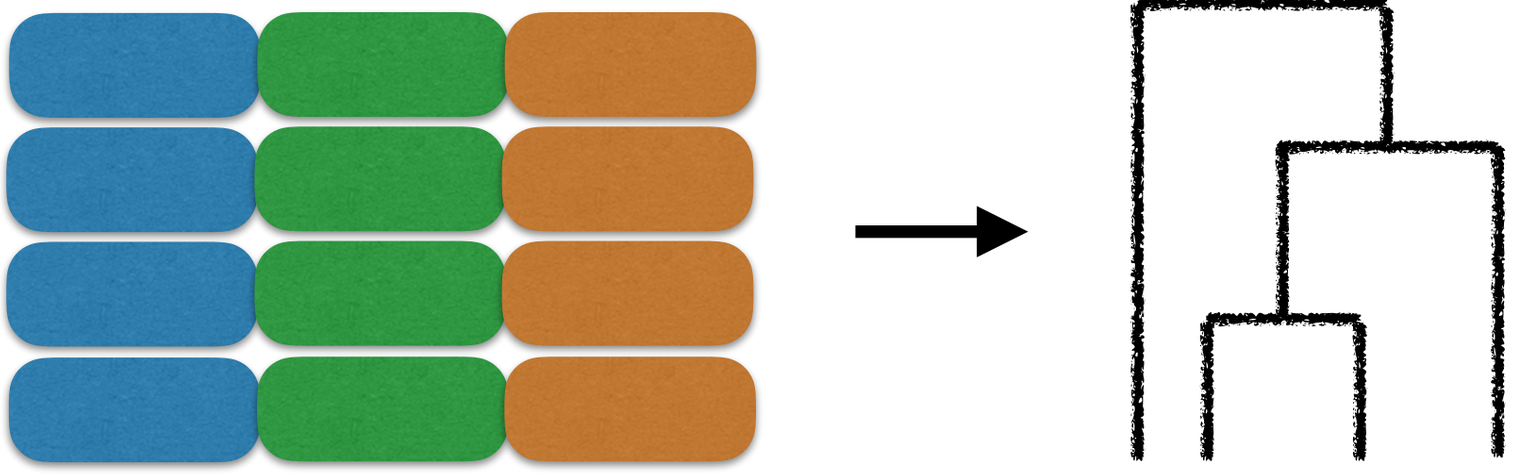


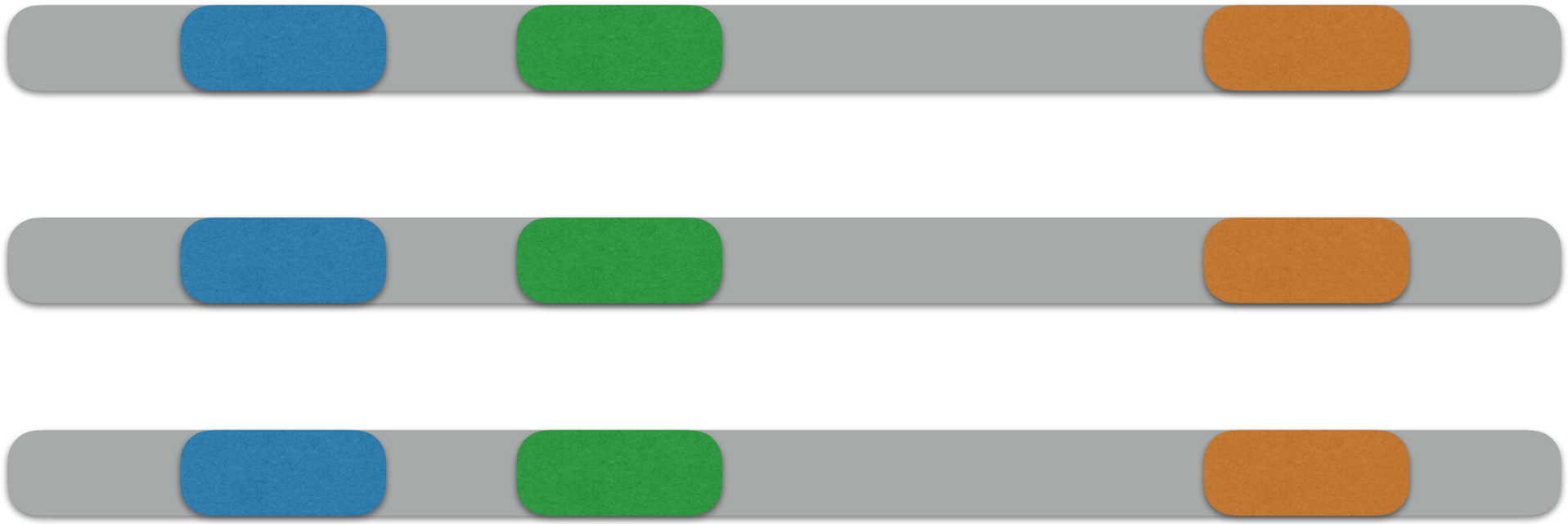
Gene
Locus
Region
Whole genome

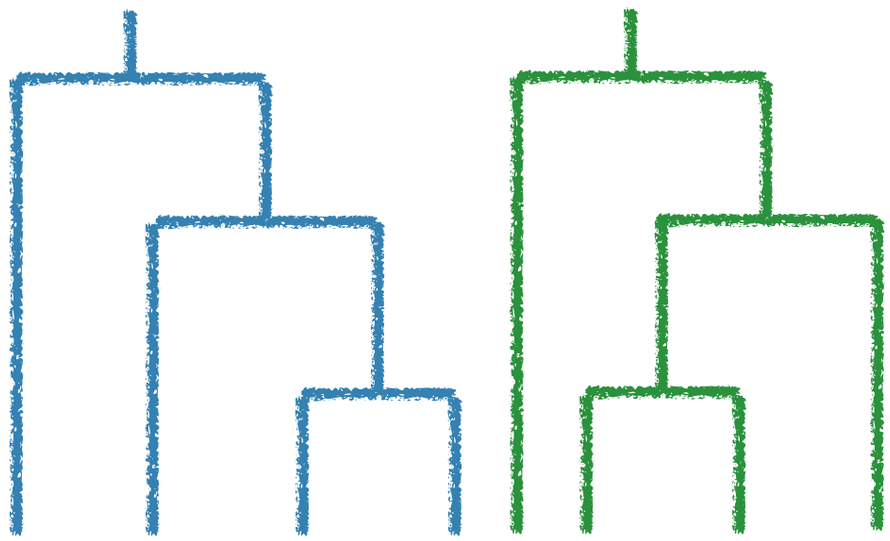
Statistically inconsistent

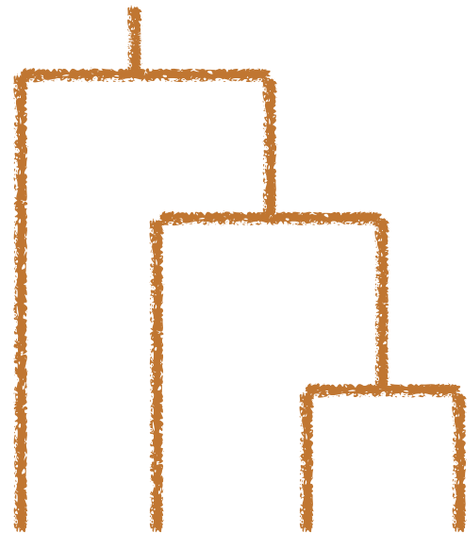
(Kubatko, Degnan, 2007)
(Roch, Steel, 2015)



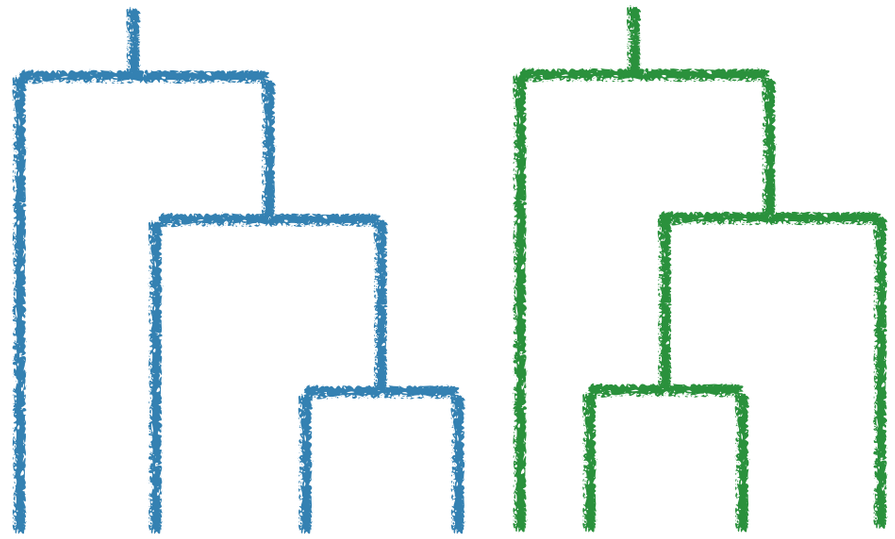
Concatenation or supermatrix

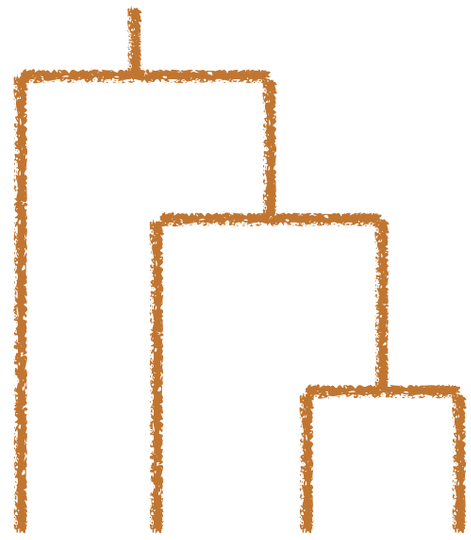






Ortholog
Recombination-free
MDL (Ané, 2011, GBE)





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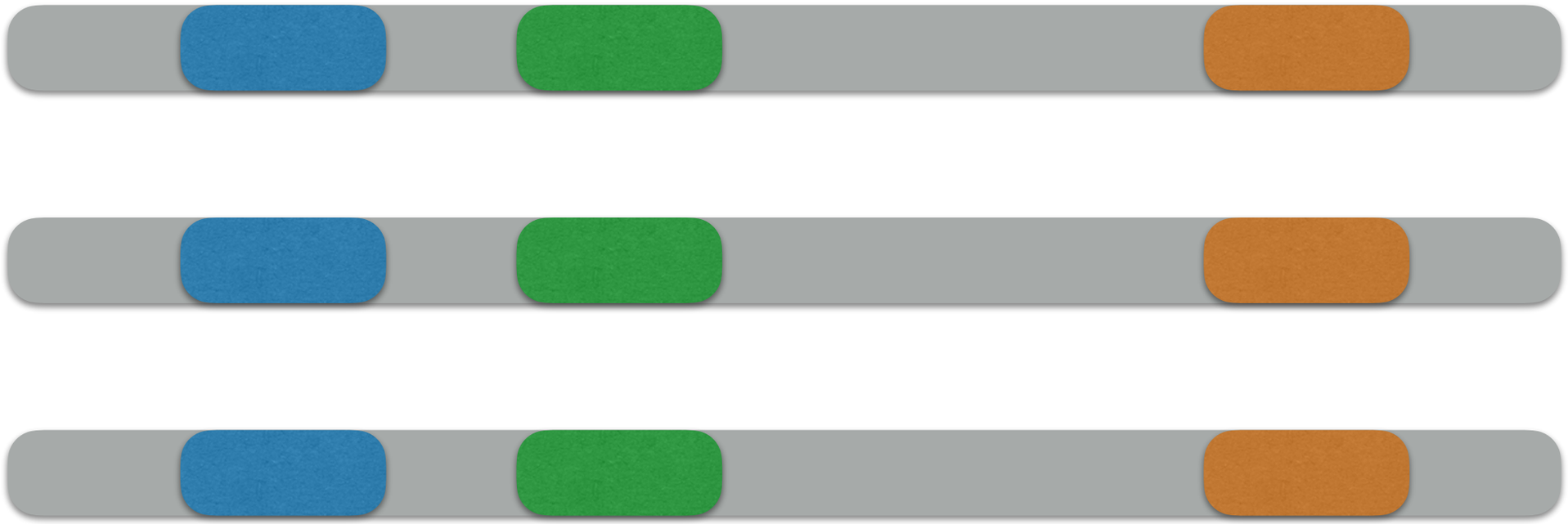
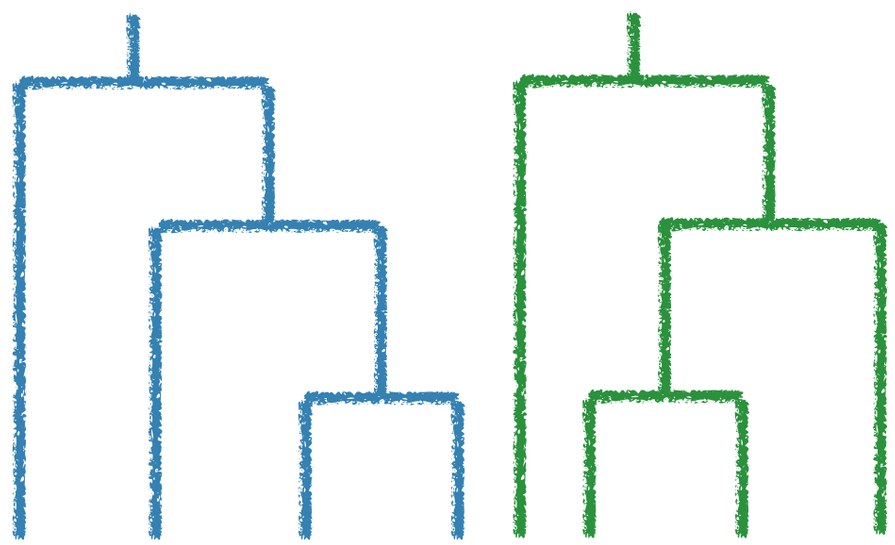
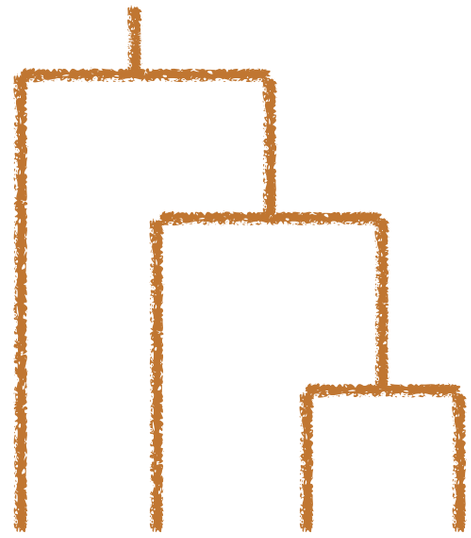


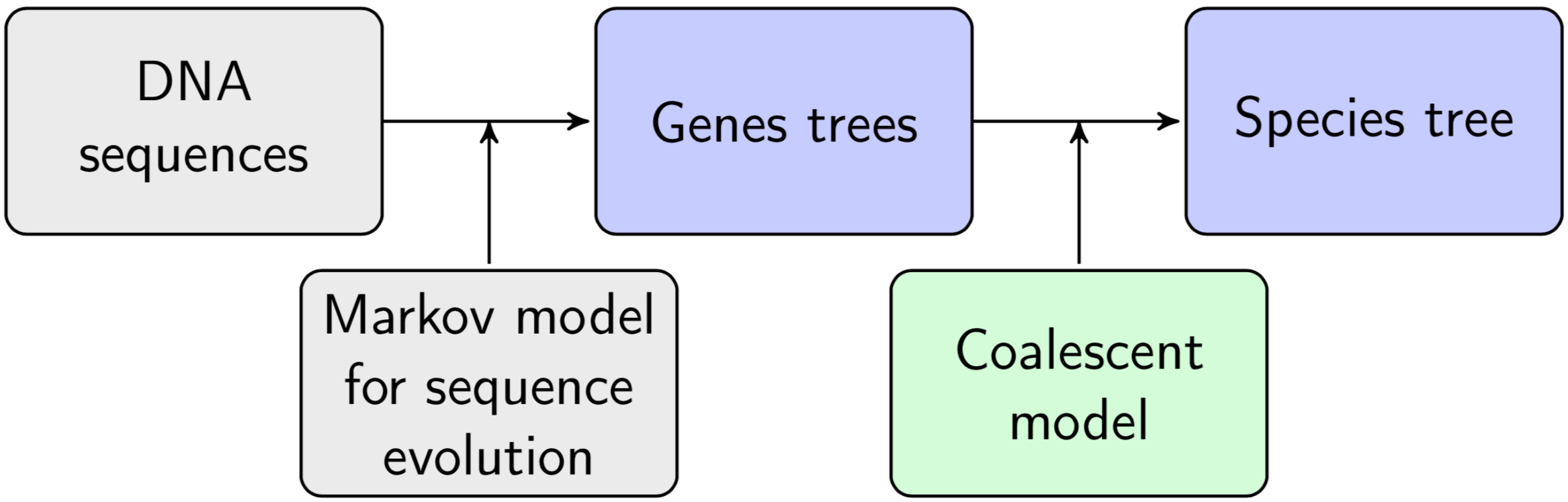
Estimate gene trees

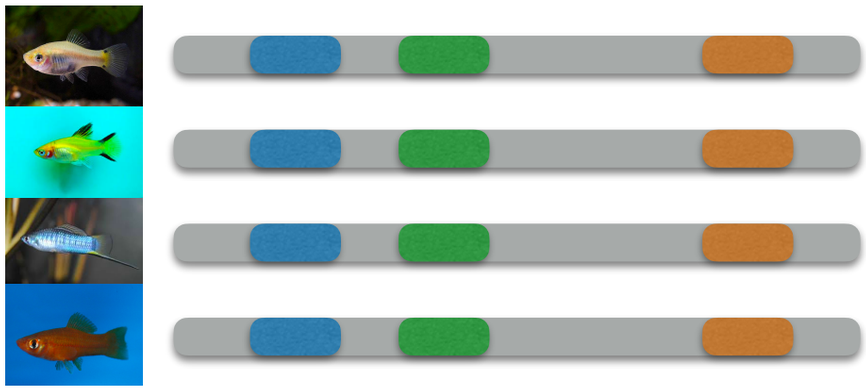
MrBayes
(Huelsenbeck, Ronquist, 2001)
RAxML
(Stamatakis, 2014)
IQ-tree 2
(Minh et al, 2020)



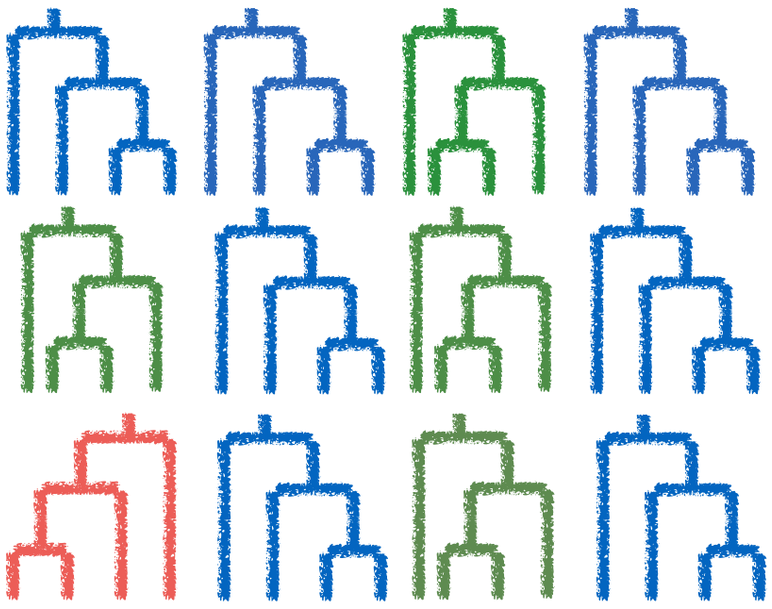


Phylogenetic inference





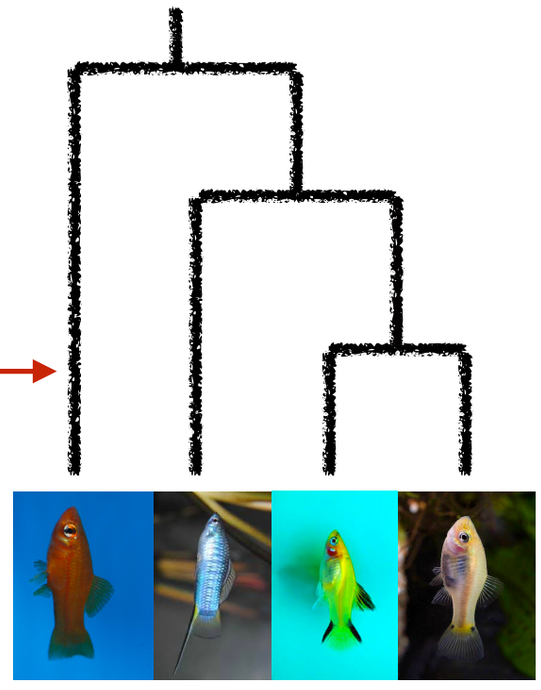
Distances
Parsimony
Likelihood
(Bayesian)

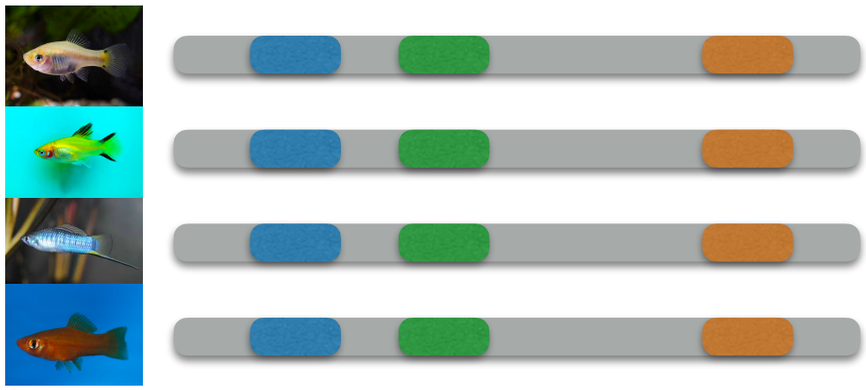


Data

$$L(T,\theta) = \prod_{i=1}^{L} P(G_i|T,\theta)$$

Max. Lik.





Distances
Parsimony
Likelihood
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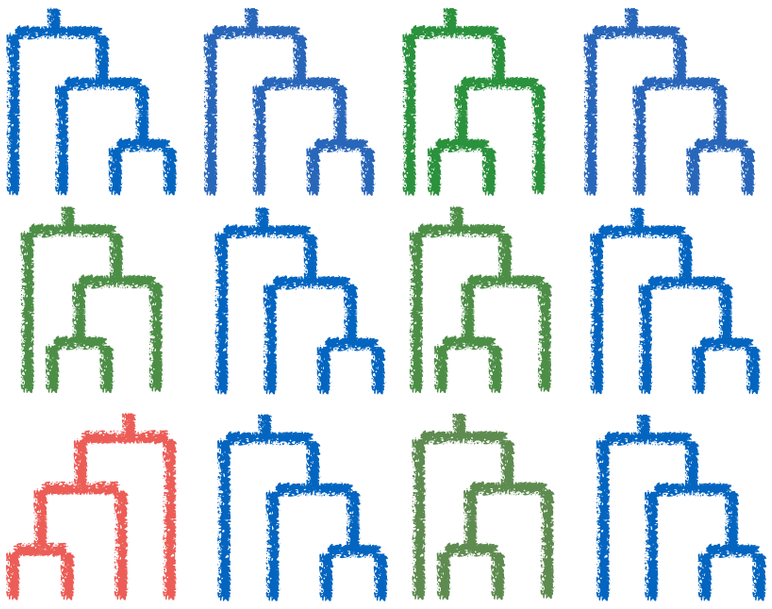

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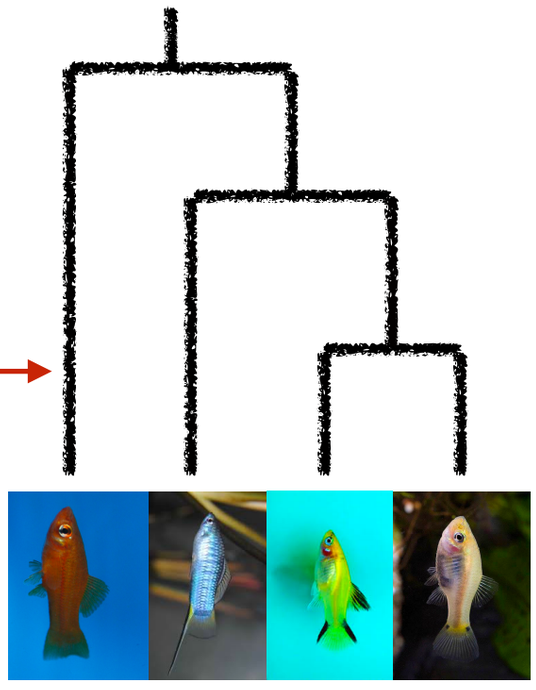
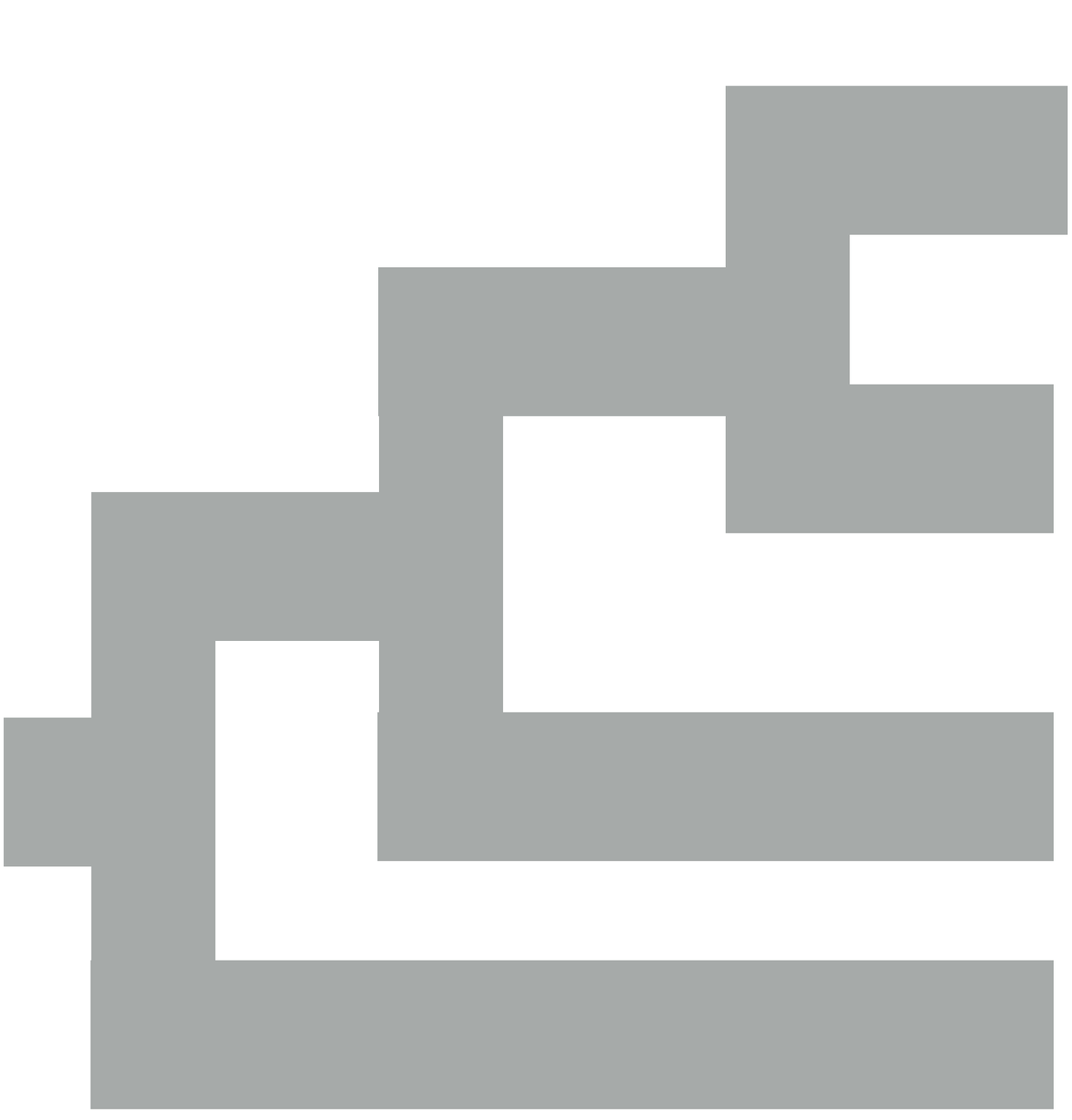
$$L(T,\theta) = \prod_{i=1}^{L} P(G_i|T,\theta)$$

Multispecies Coalescent Model

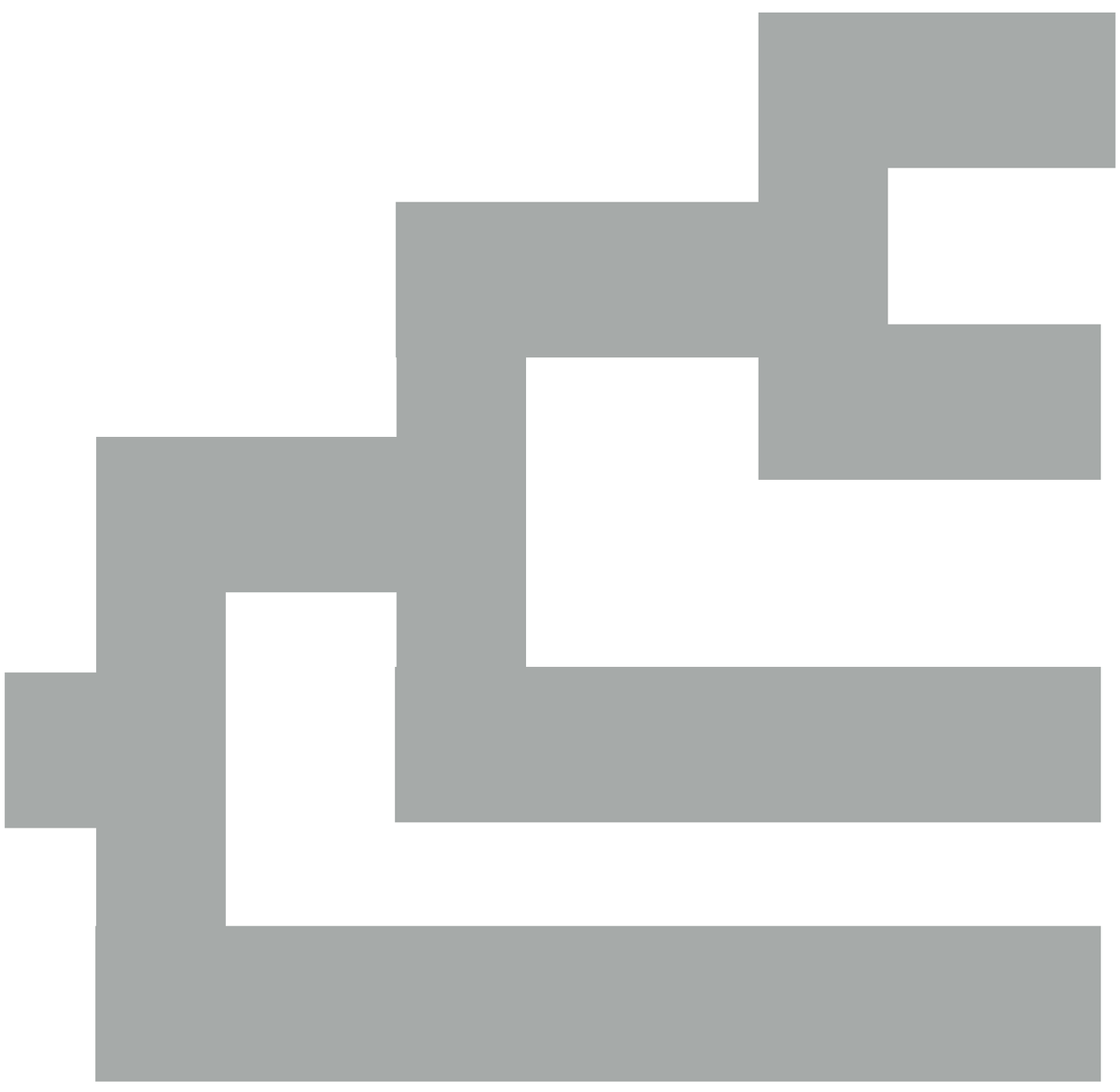
Max. Lik.



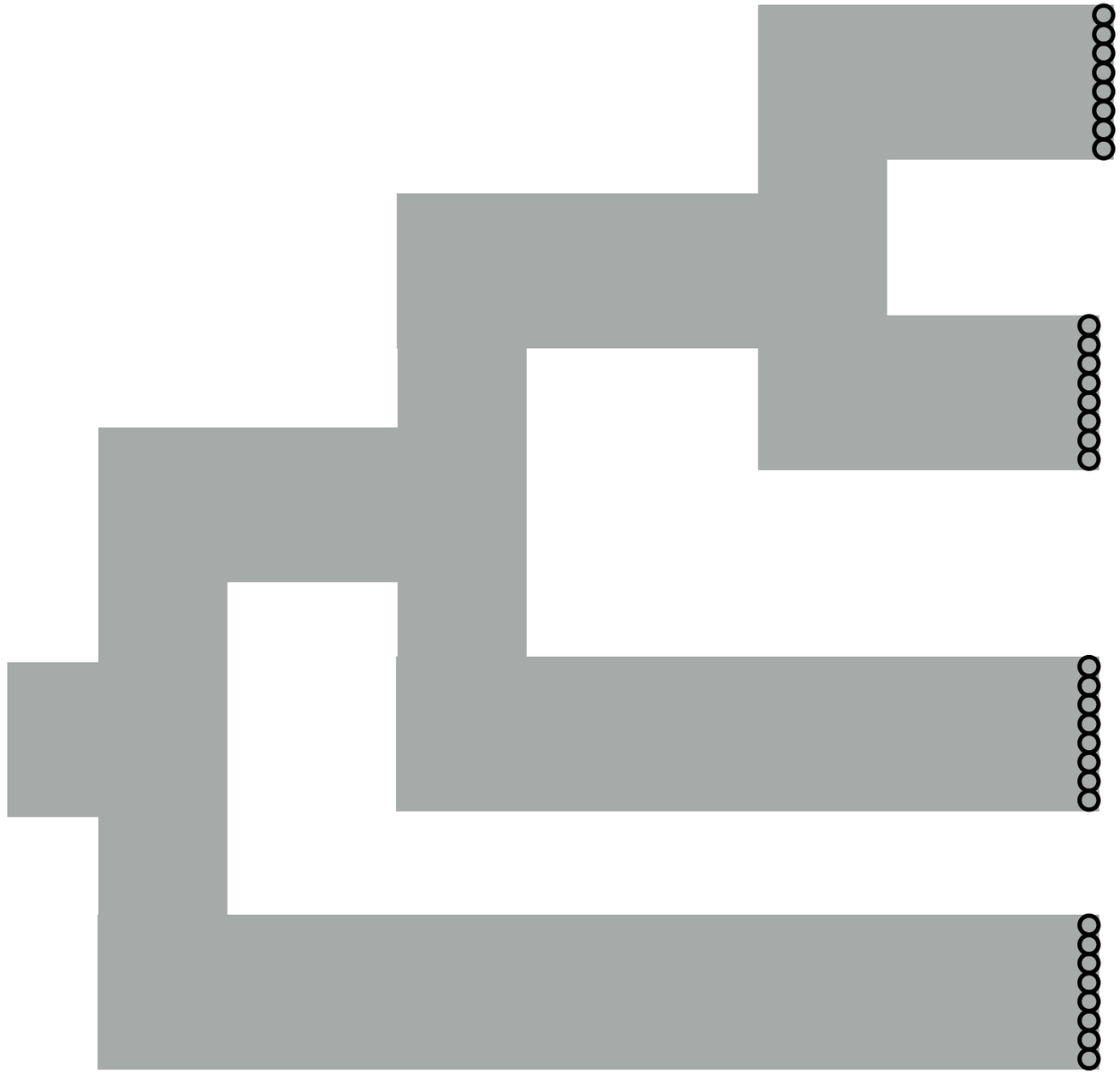
Coalescent model

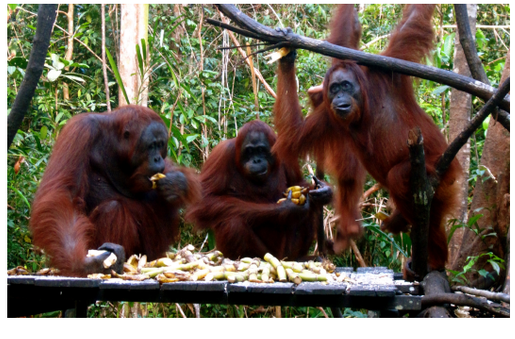


Coalescent model



Coalescent model





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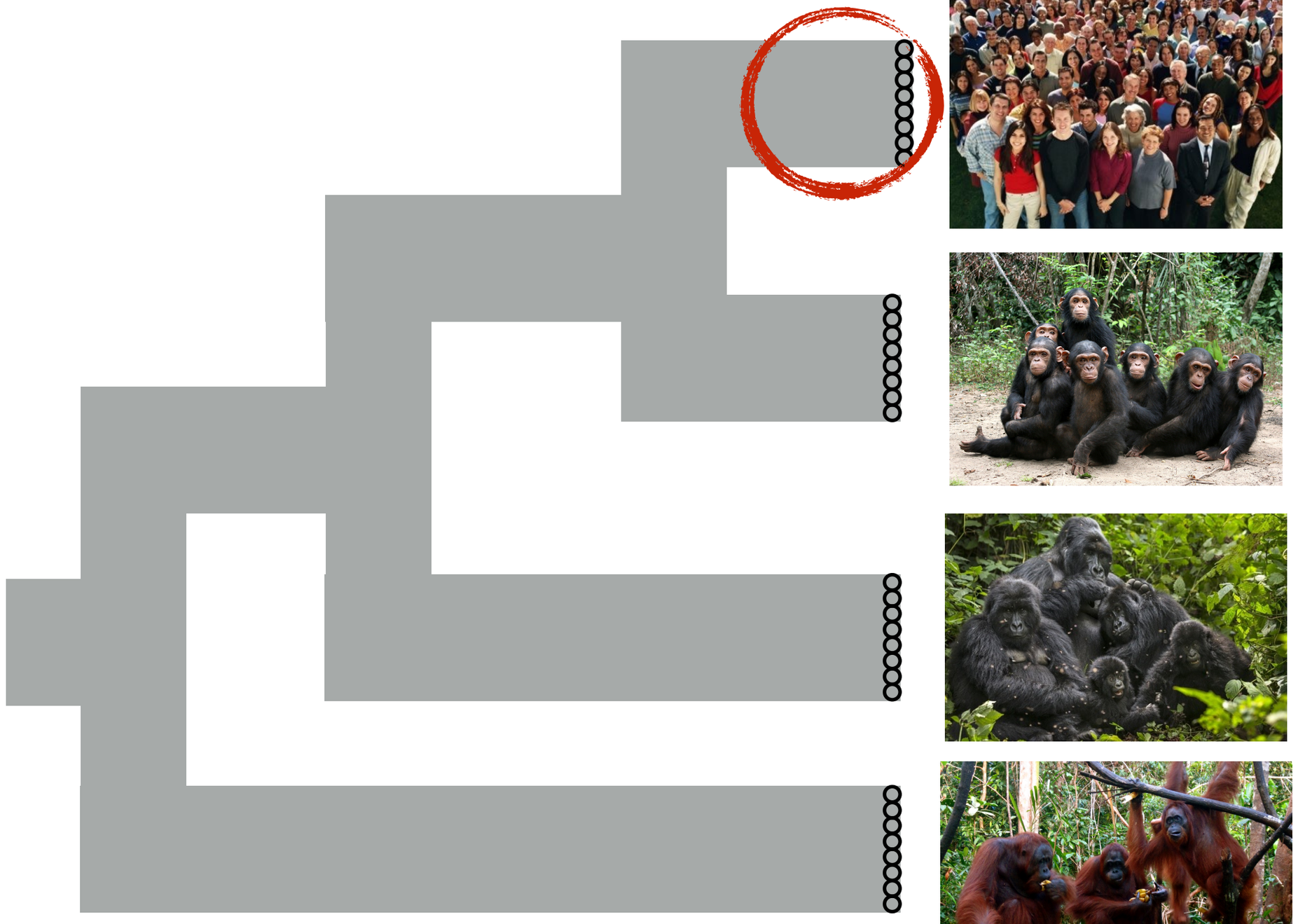
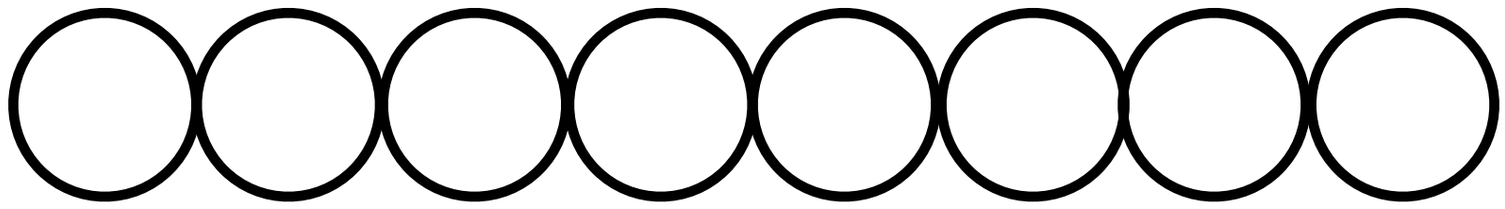
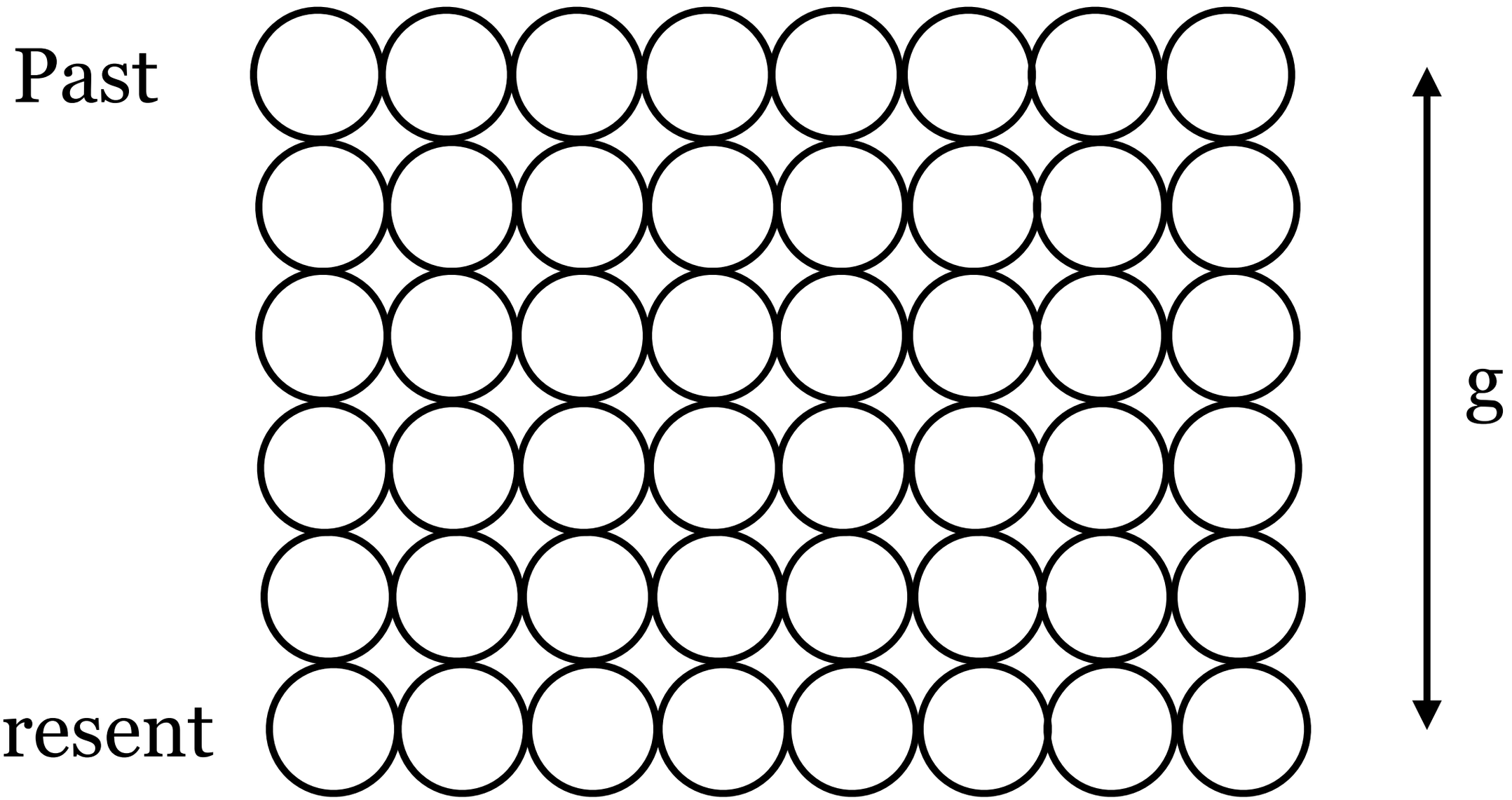


Coalescent model within 1 population

Present 

Coalescent model within 1 population



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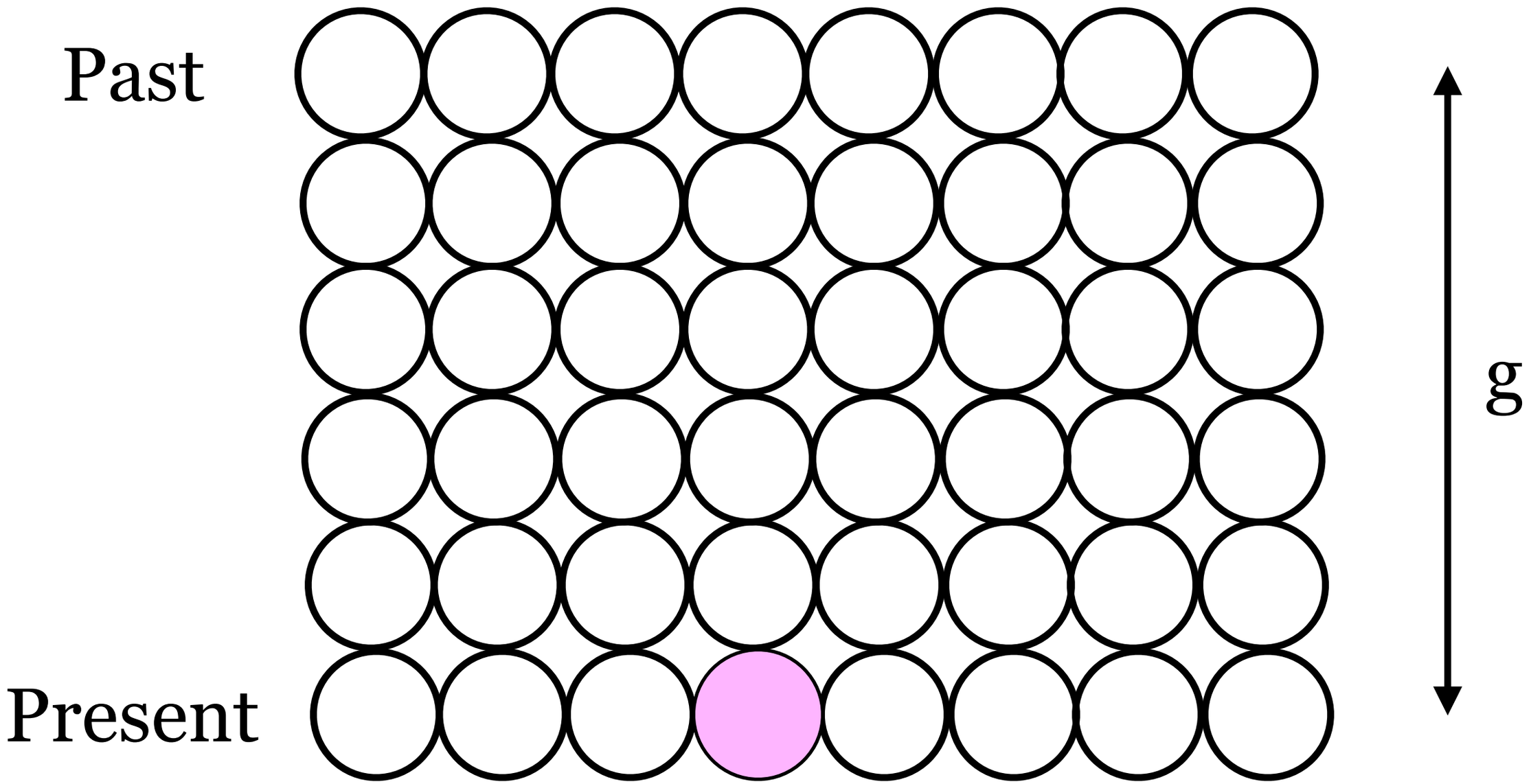

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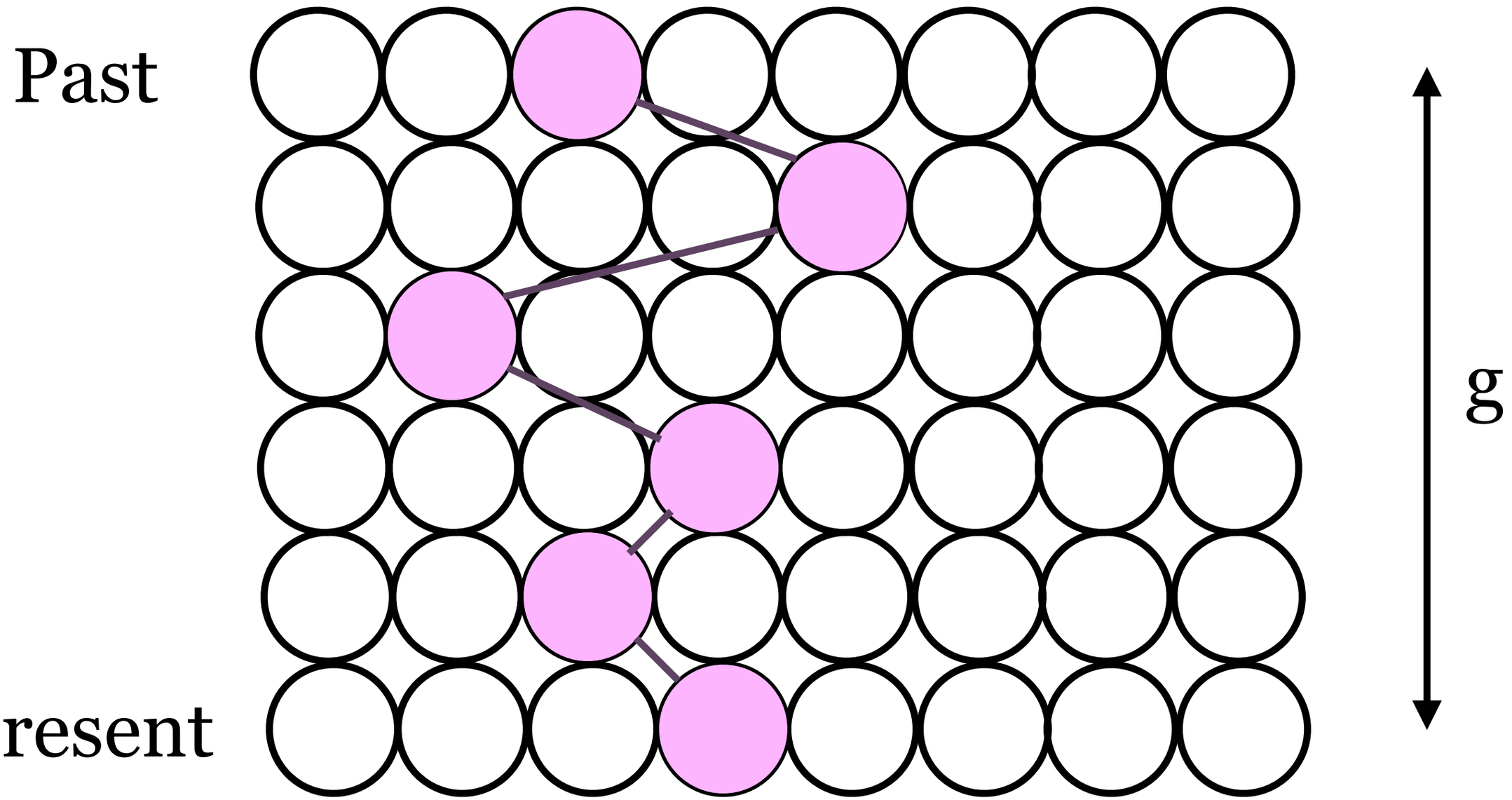
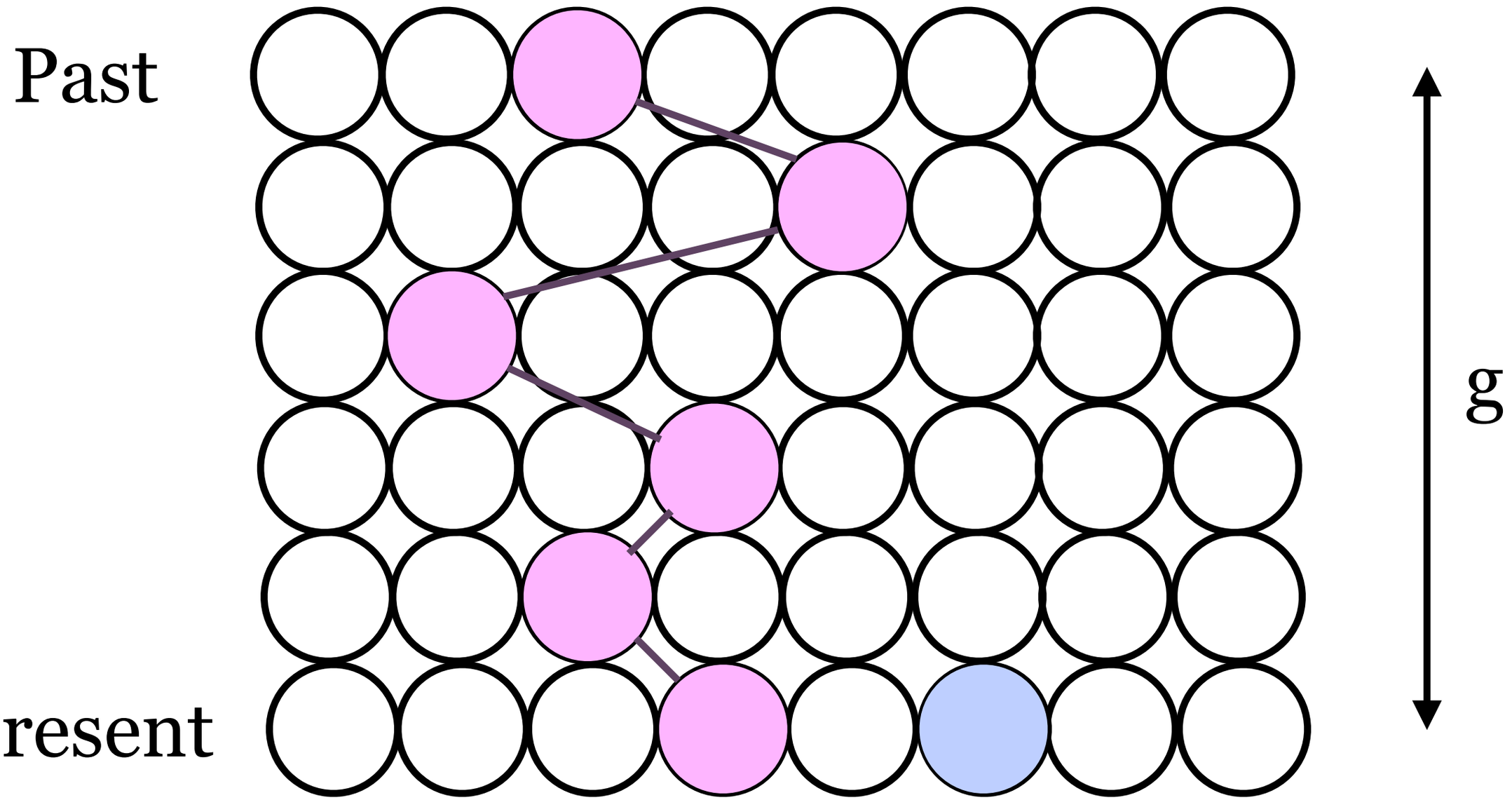


No selection: uniform probability 1/N

Coalescent model within 1 population



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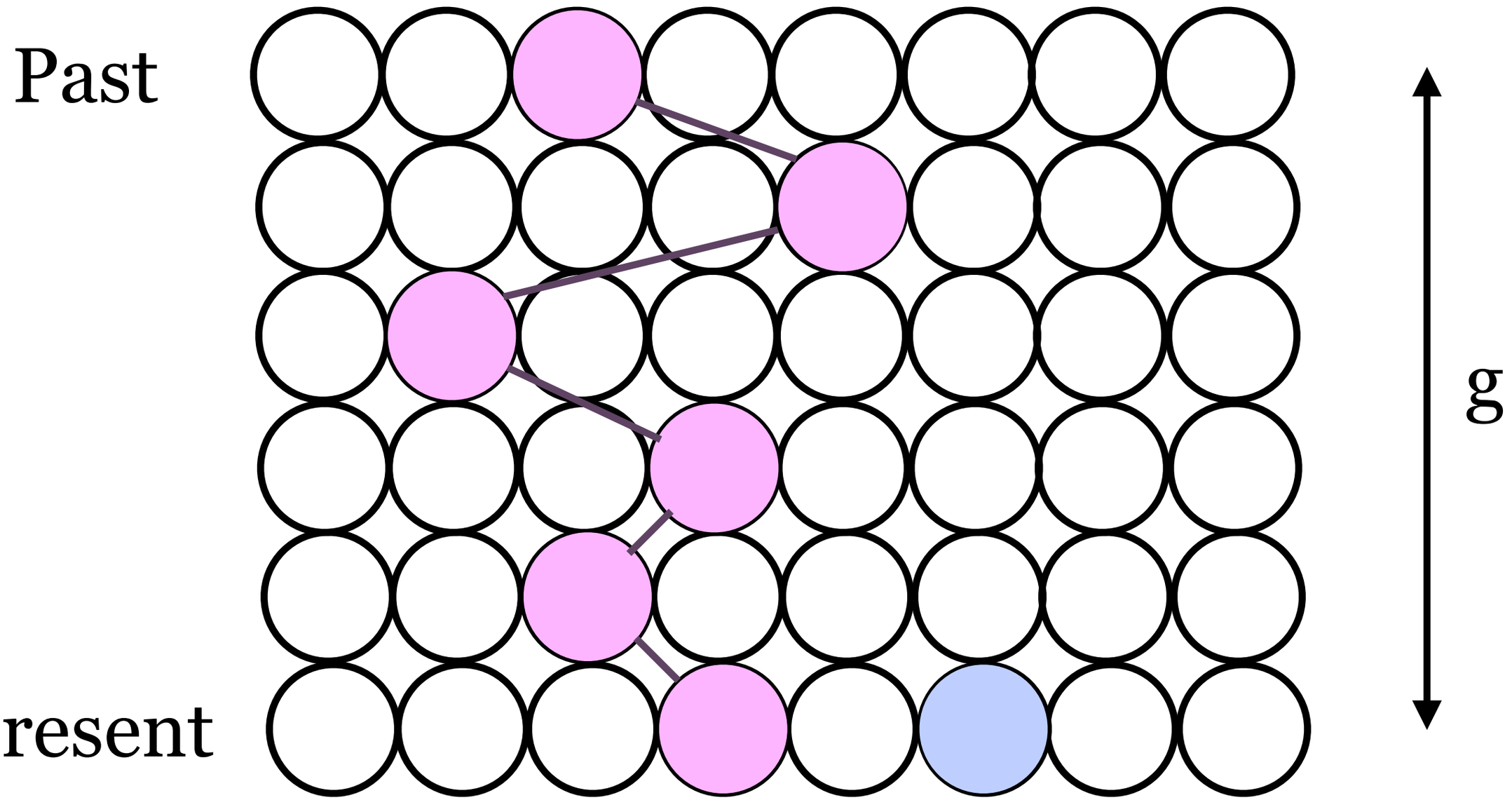
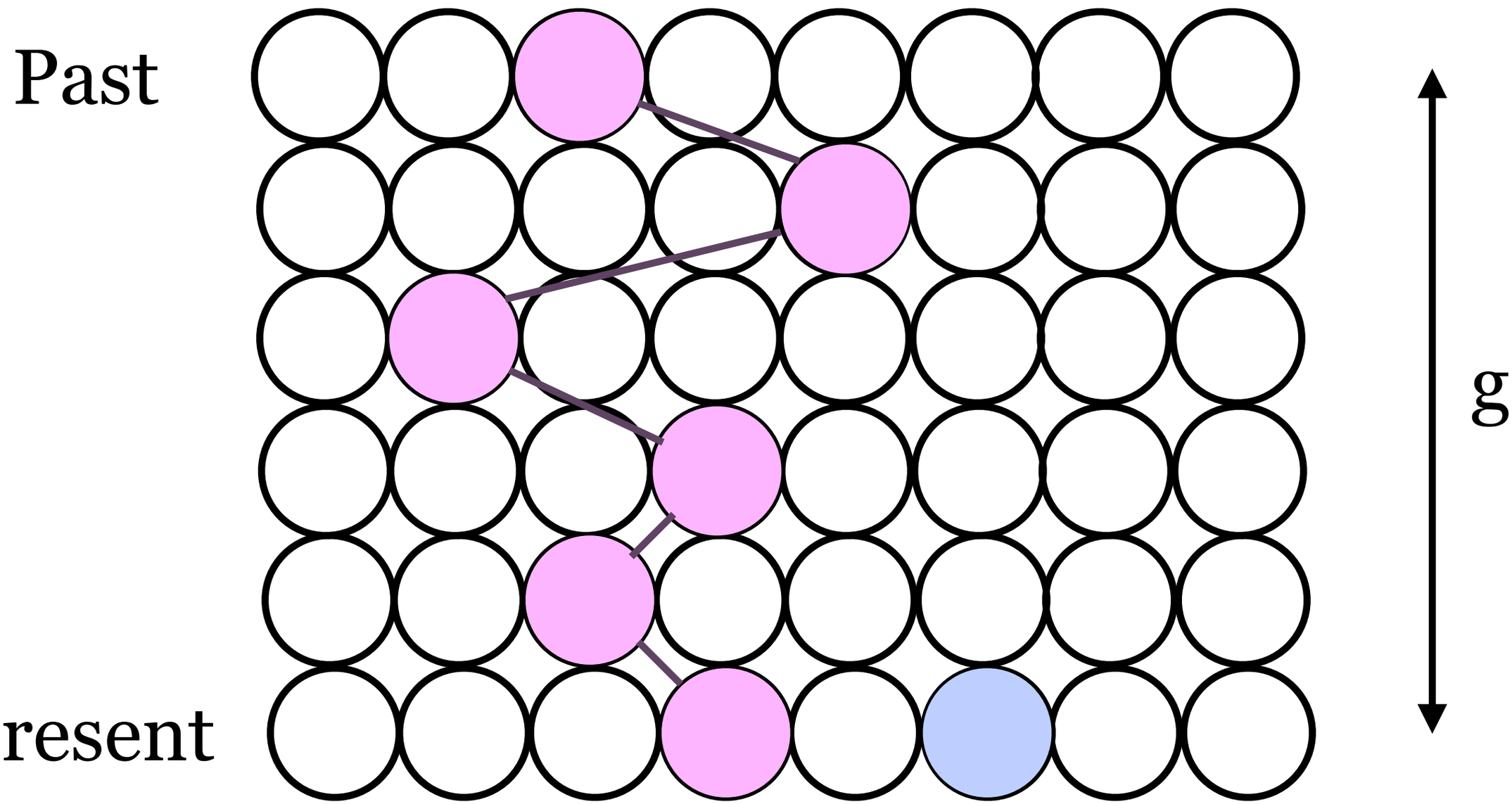


How many generations do we have to wait for these two individuals to reach a common ancestor?

Coalescent model within 1 population

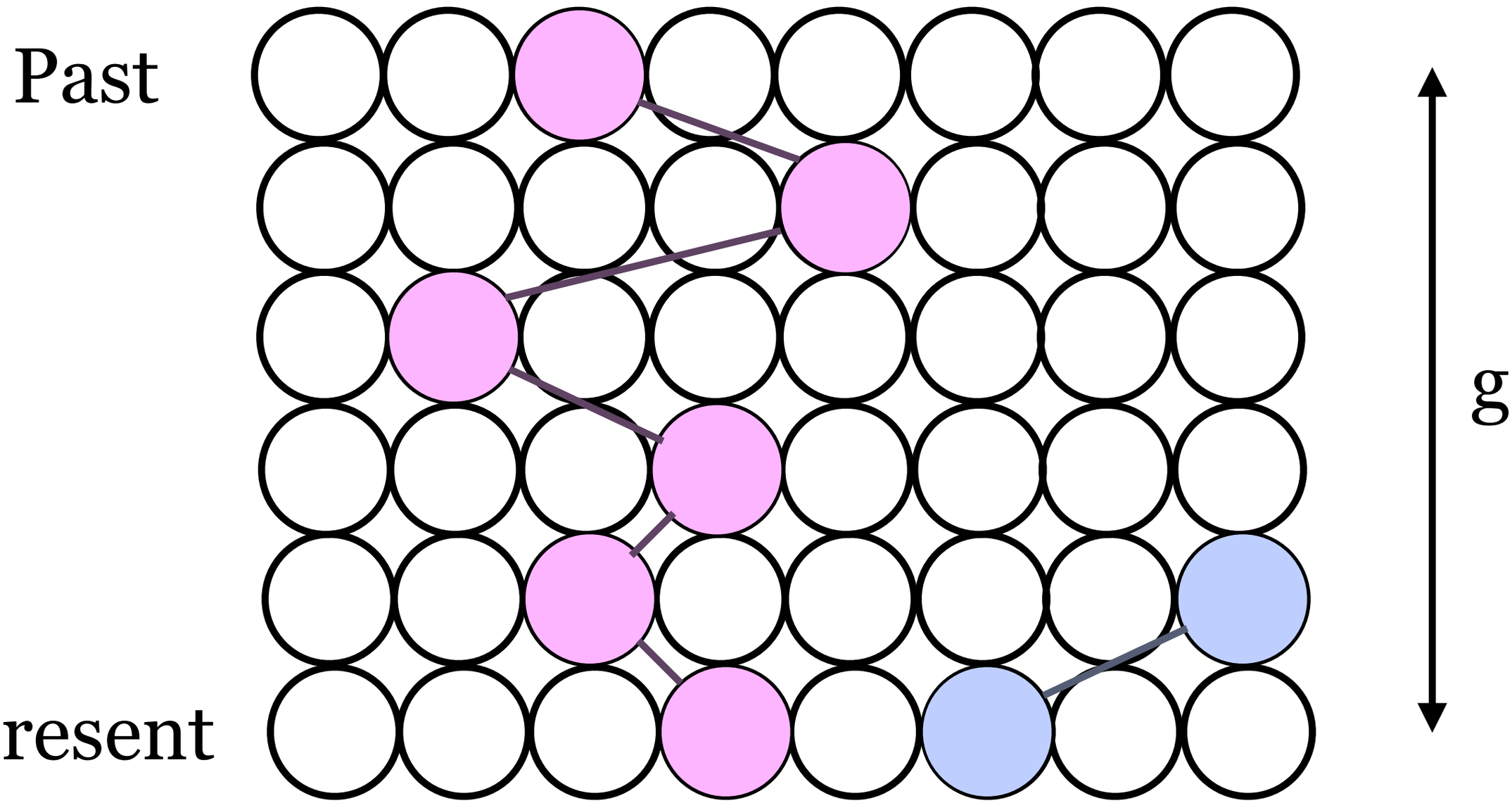


How many generations do we have to wait for these two individuals to reach a common ancestor?

$$P(\text{coalesce}) = \frac{1}{N}$$

$$P(\text{no coalesce}) = 1 - \frac{1}{N}$$

Coalescent model within 1 population



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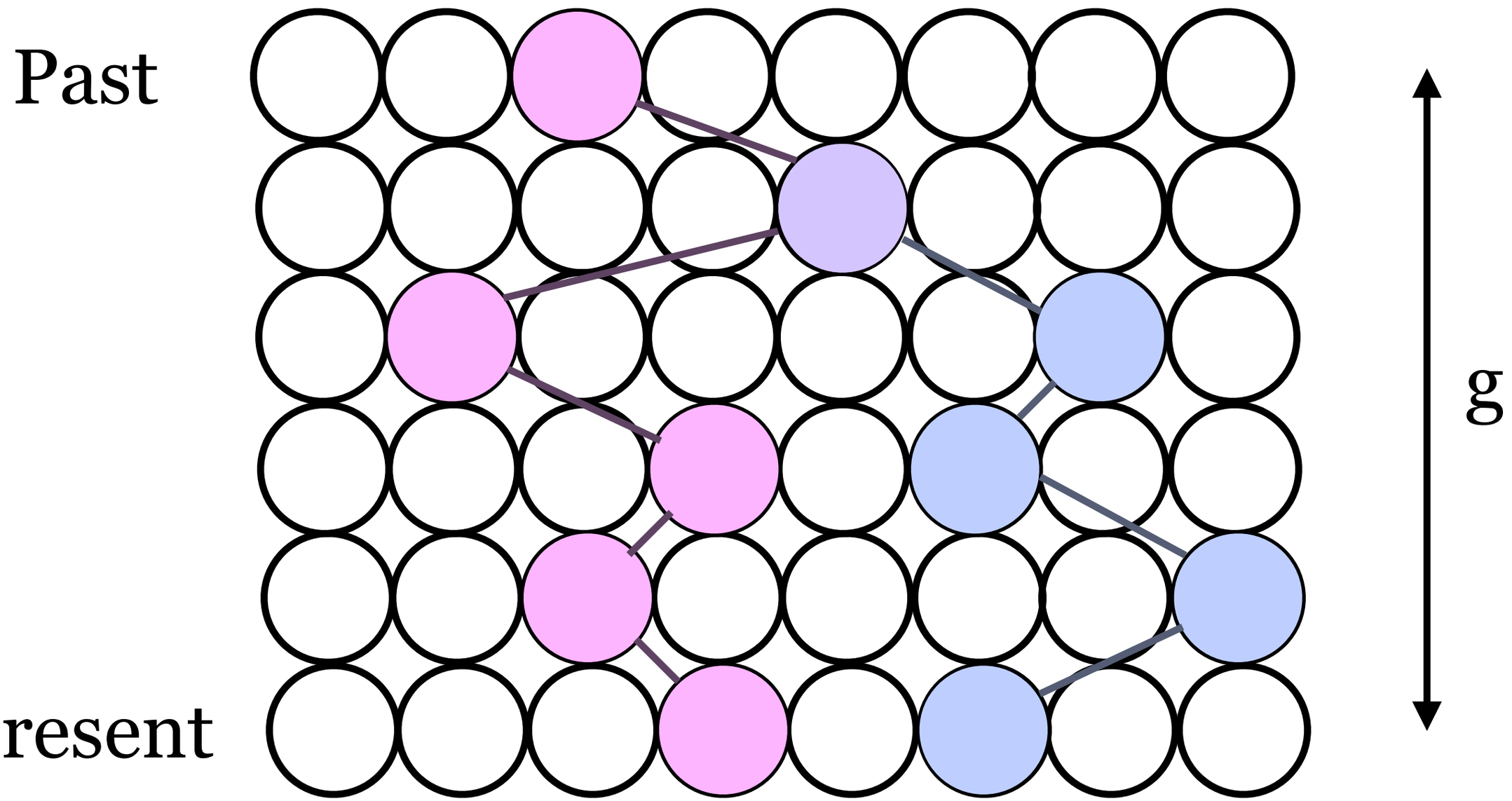
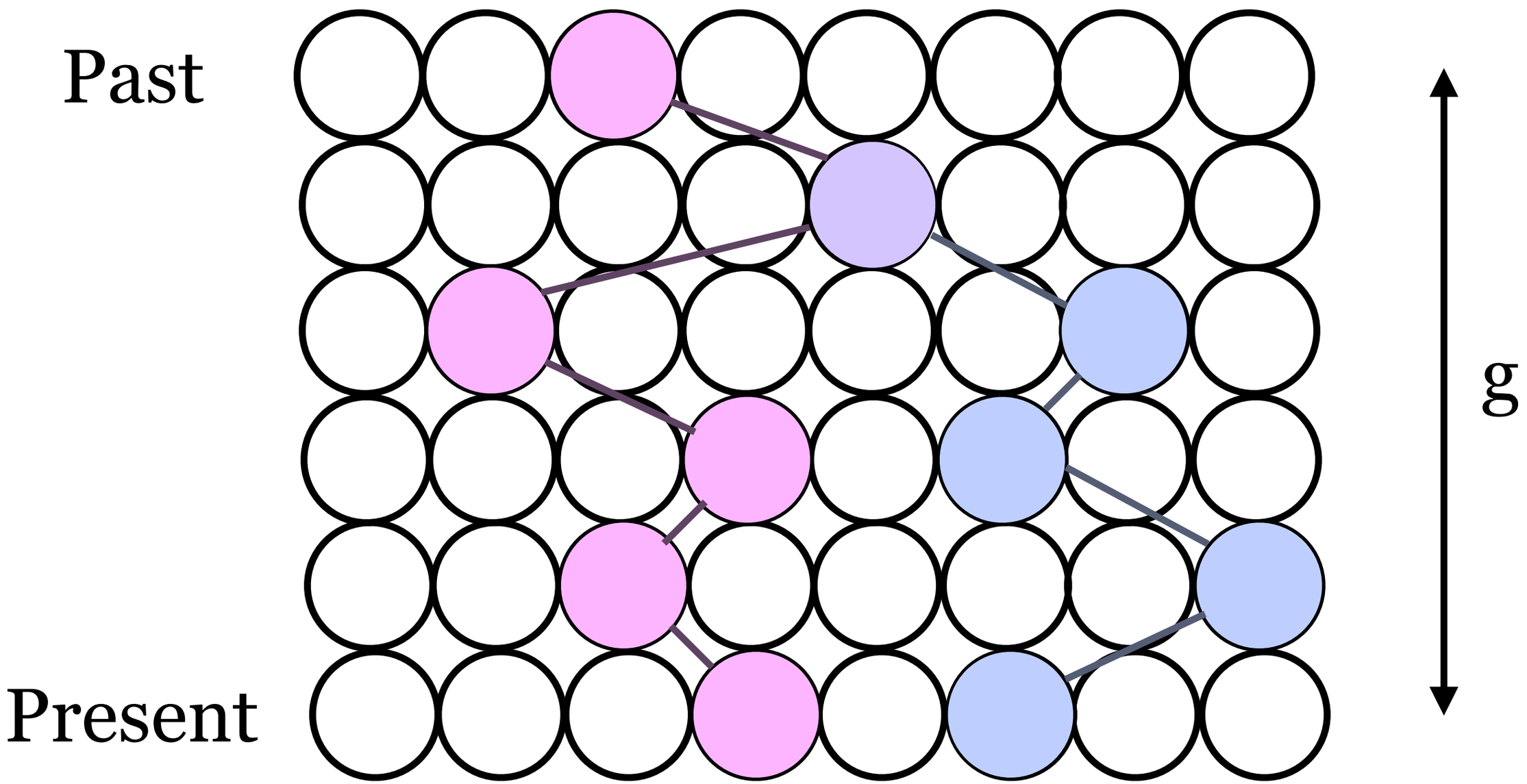
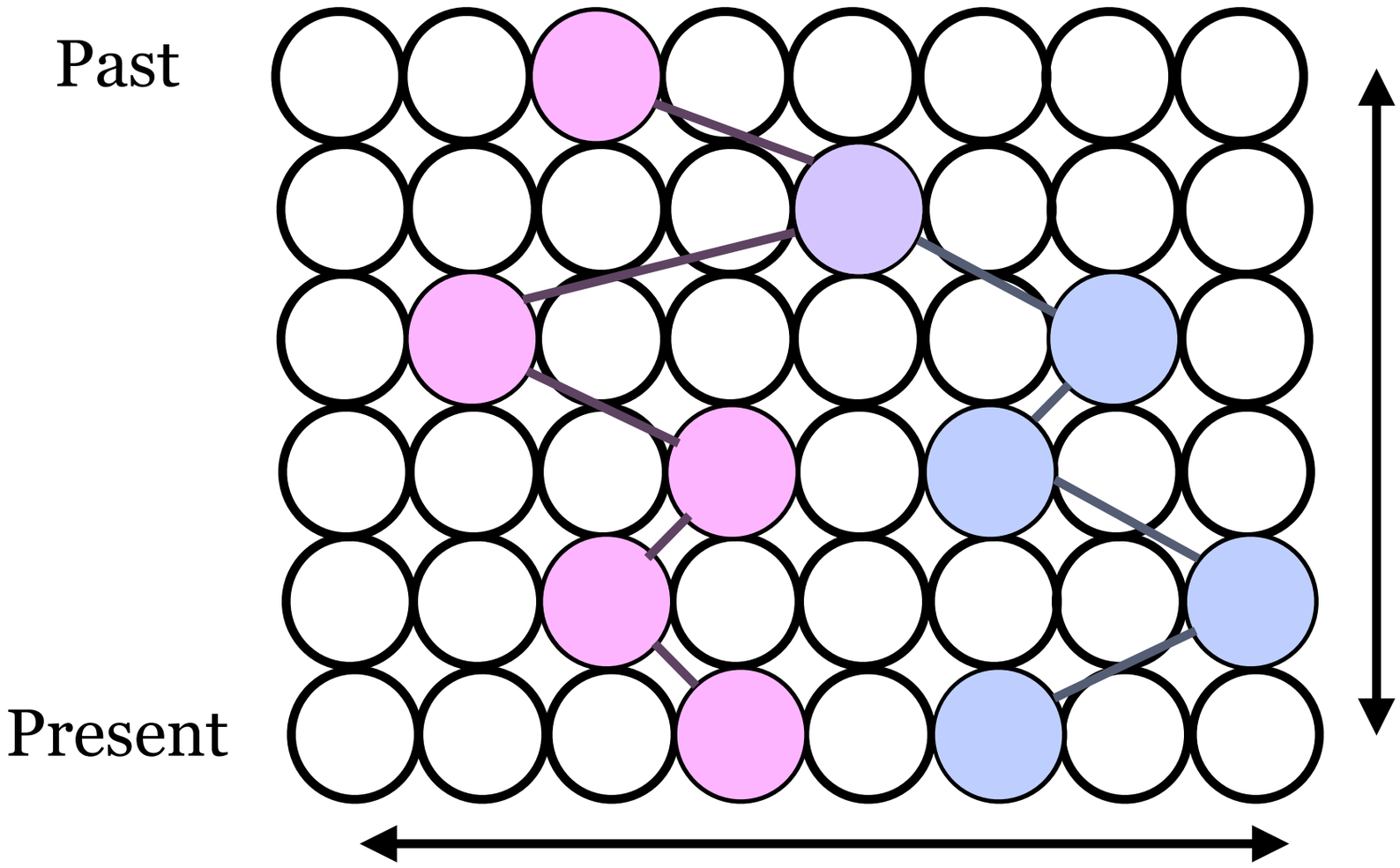


Coalescent model within 1 population



Coalescent event

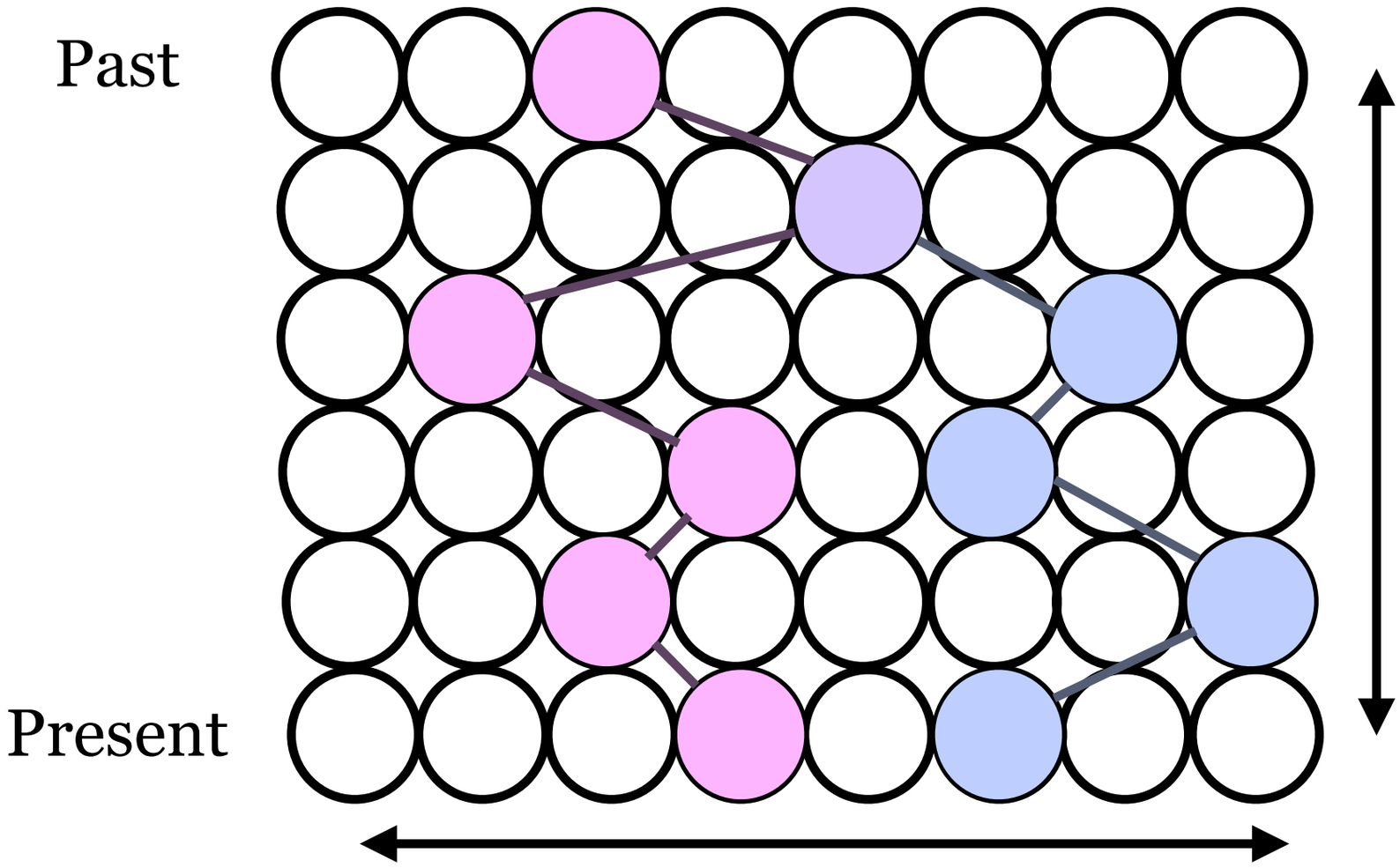
Coalescent model within 1 population



Probability of no coalescence in g generations: $\left(1-\frac{1}{N}\right)^{g}$

$$t = g/N \Rightarrow \left(1-\frac{t}{Nt}\right)^{Nt} \xrightarrow[N\to\infty]{} e^{-t}$$

Coalescent model within 1 population



- Haploid population of constant size (N)
- No selection: uniform probability 1/N

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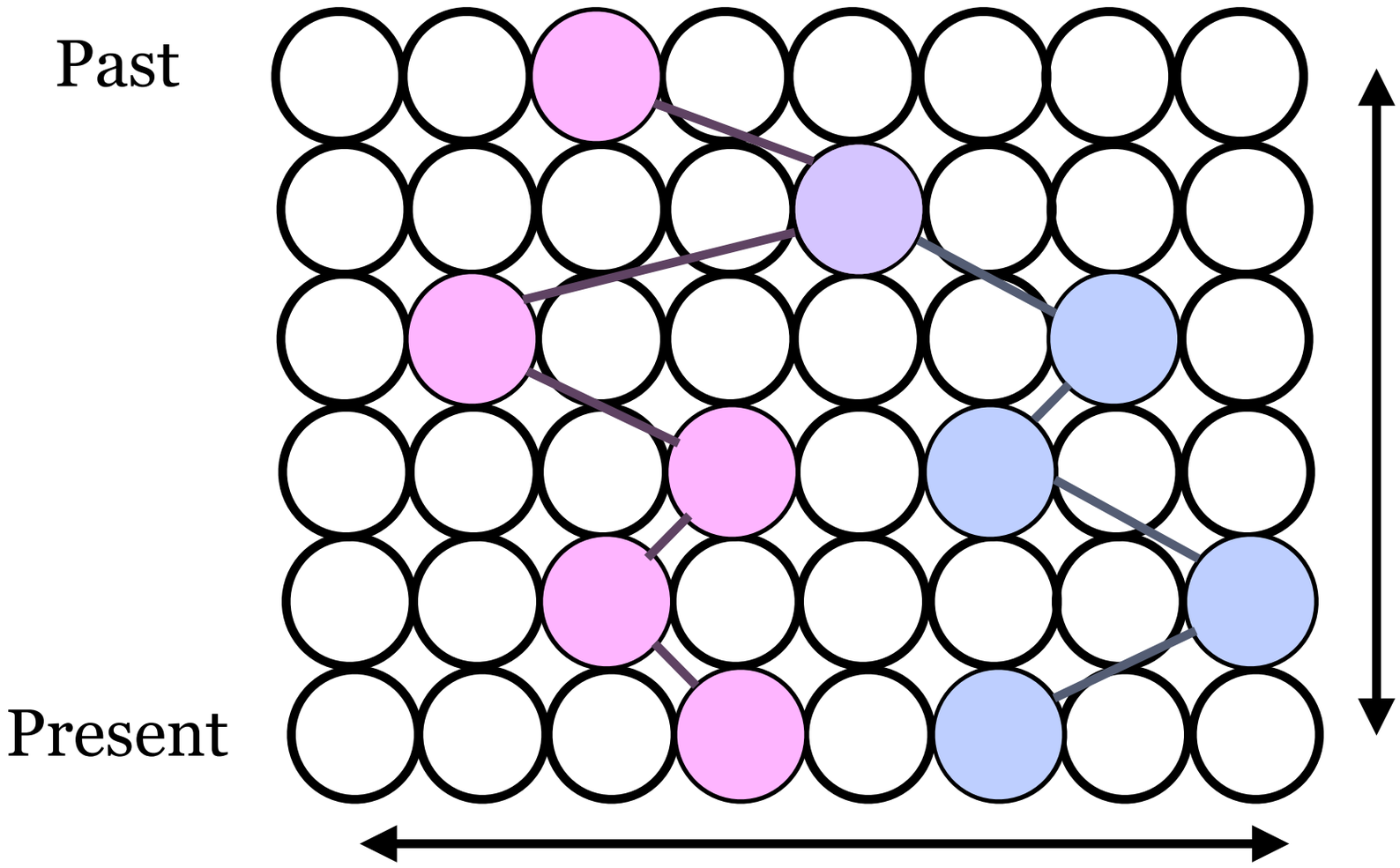
Coalescent model within 1 population



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Multispecies coalescent on a tree

Probability of no coalescence in g generations: $\left(1-\frac{1}{N}\right)^{g}$

$$t = g/N \Rightarrow \left(1-\frac{t}{Nt}\right)^{Nt} \xrightarrow[N\to\infty]{} e^{-t}$$

$$T = \frac{g}{N} \text{ coalescent units } \sim Exp(1)$$

Time to coalesce

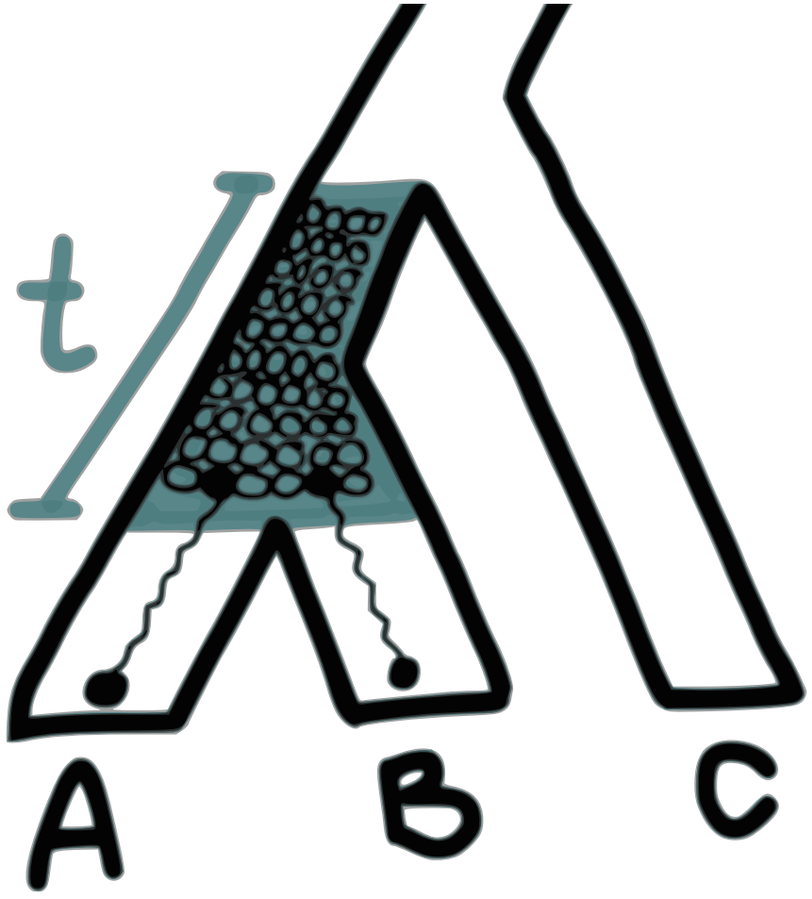
$$P(T > t) = e^{-t}$$

Probability of no coalescence in time t

Multispecies coalescent on a tree

Probability of no coalescence in g generations: $\left(1-\frac{1}{N}\right)^{g}$

$$t = g/N \Rightarrow \left(1-\frac{t}{Nt}\right)^{Nt} \xrightarrow[N\to\infty]{} e^{-t}$$



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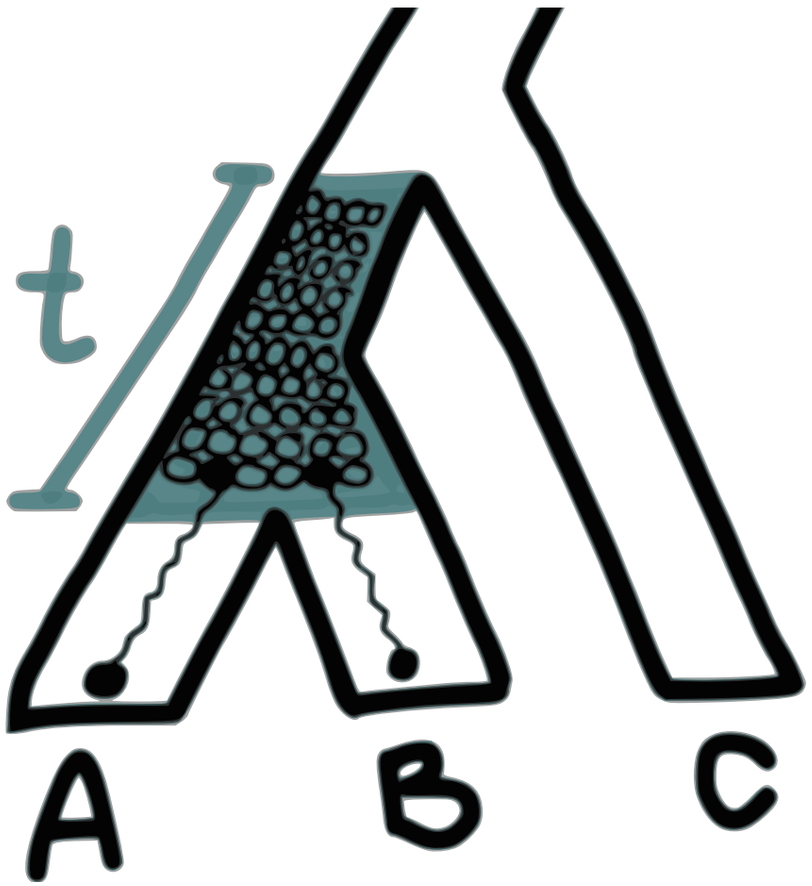
Multispecies coalescent on a tree

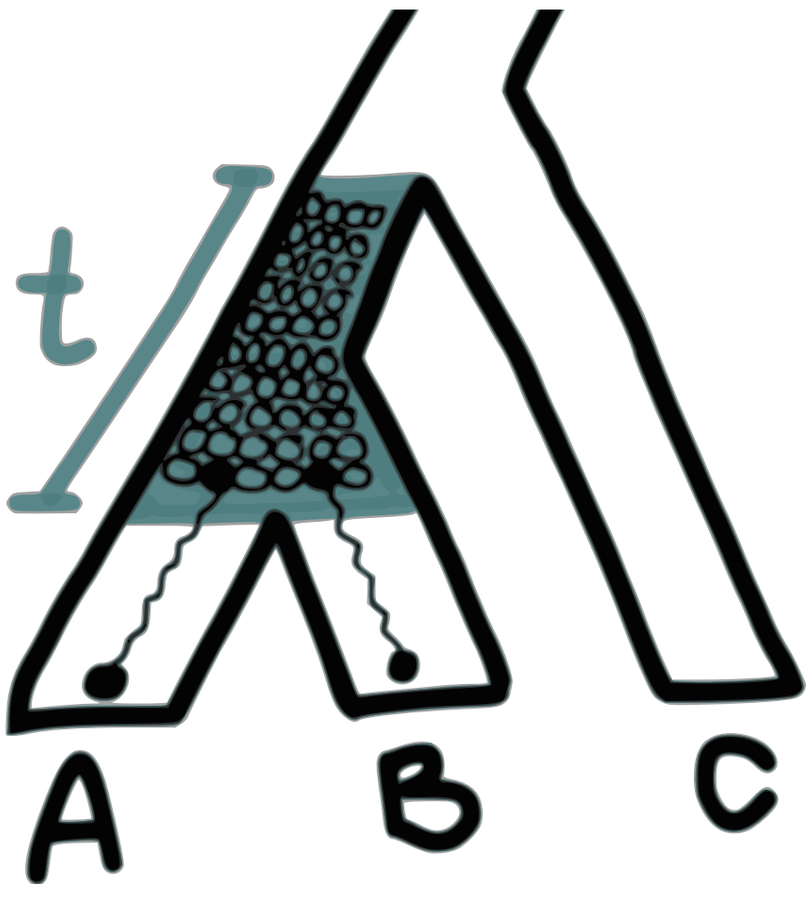


$$\mathbb{P}\left(\begin{array}{ccc} & \wedge & \\ A & B & C \end{array}\right) =$$

$$P(T > t) = e^{-t}$$

Probability of no coalescence in time t

Multispecies coalescent on a tree

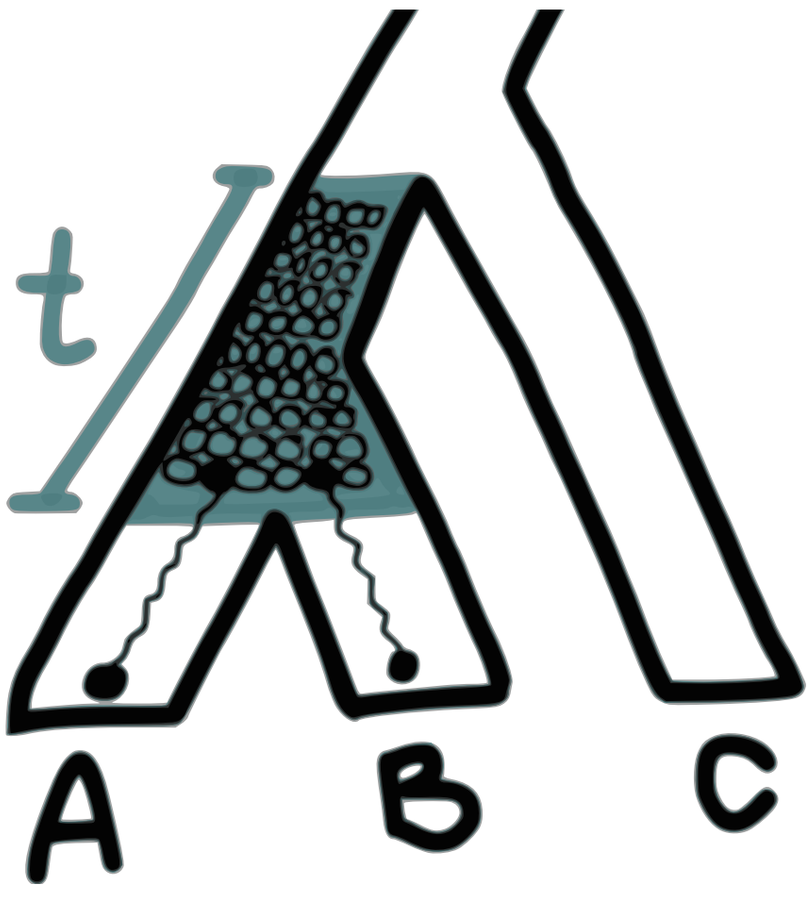


$$\mathbb{P}\left(\begin{array}{c}\\ \wedge \\ \text{A B C}\end{array}\right) = 1 - e^{-t}$$

$$P(T > t) = e^{-t}$$

Probability of no coalescence in time t

Multispecies coalescent on a tree



$$\mathbb{P}\left(\begin{array}{c}\\ \wedge \\ \text{A B C}\end{array}\right) =$$

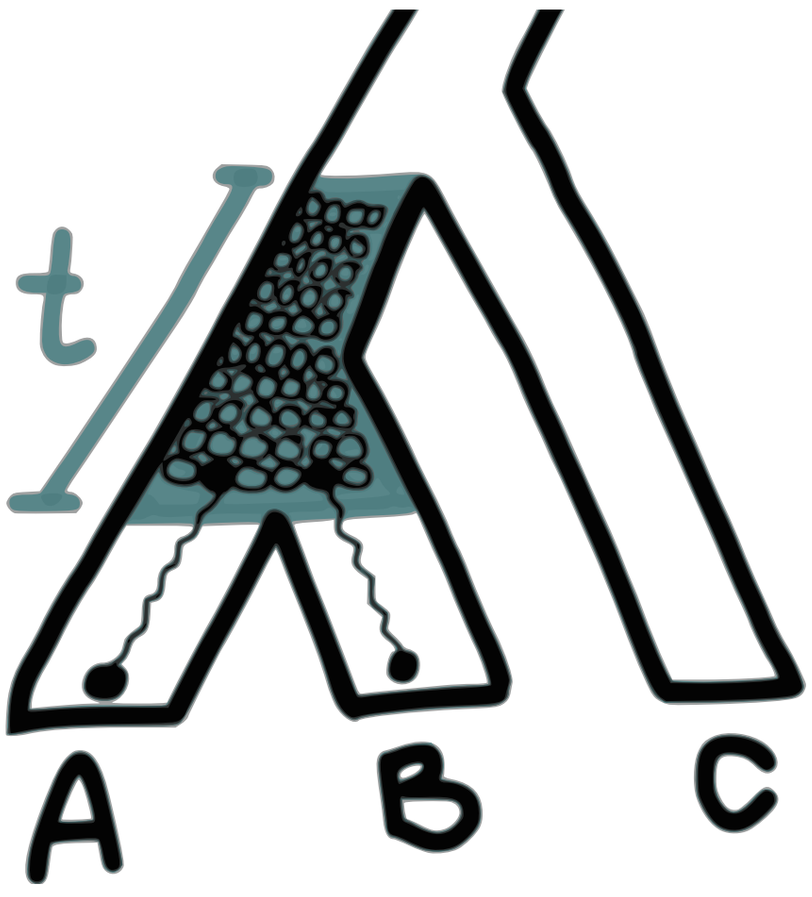
$$1 - e^{-t}$$

$$+$$

$$P(T > t) = e^{-t}$$

Probability of no coalescence in time t

Multispecies coalescent on a tree

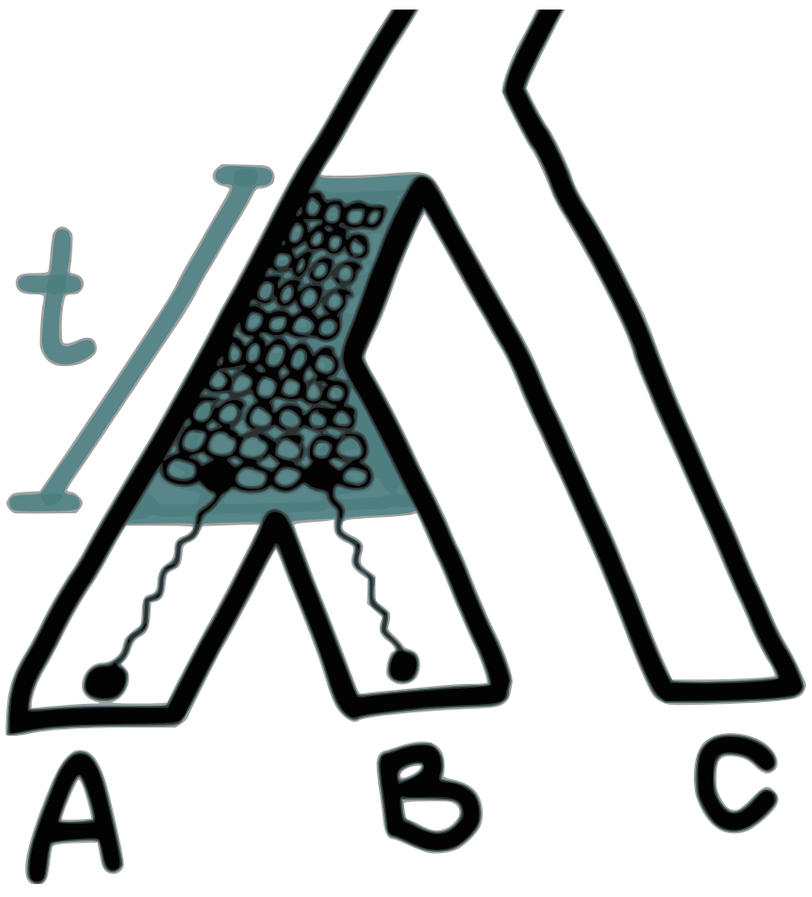


$$\mathbb{P}\left(\begin{array}{c}\\ \wedge \\ \text{A B C}\end{array}\right) = 1 - e^{-t} + e^{-t} \times 1/3$$

$$P(T > t) = e^{-t}$$

Probability of no coalescence in time t

Multispecies coalescent on a tree



$$P(T > t) = e^{-t}$$

Probability of no coalescence in time t

$$\mathbb{P}\left(\begin{array}{ccc} & \wedge & \\ A & B & C \end{array}\right) =$$

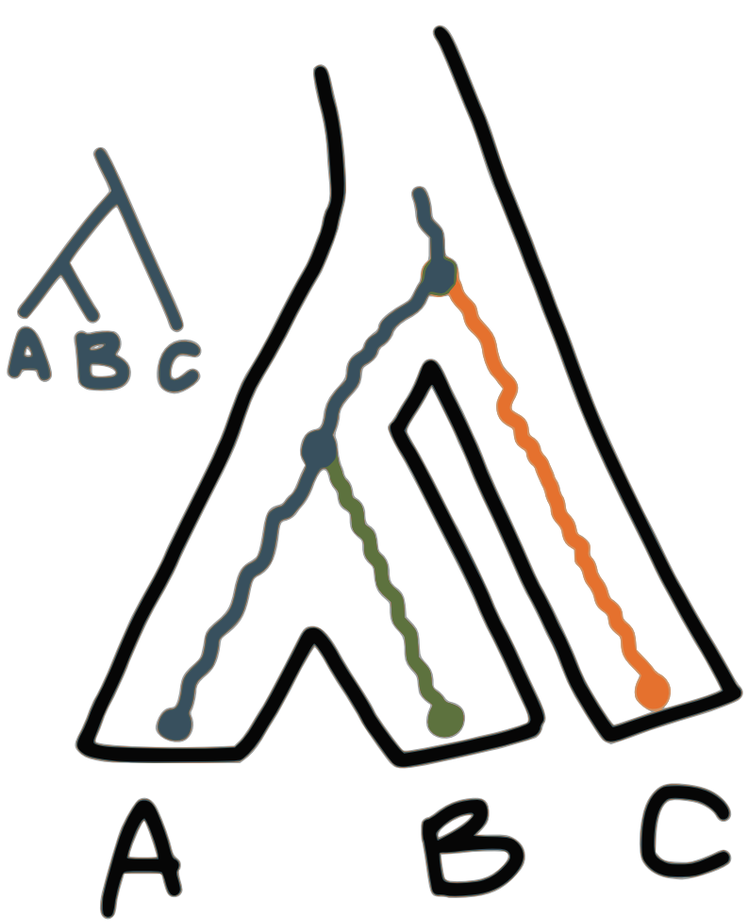
$$1 - e^{-t}$$

$$+$$

$$e^{-t} \times 1/3$$

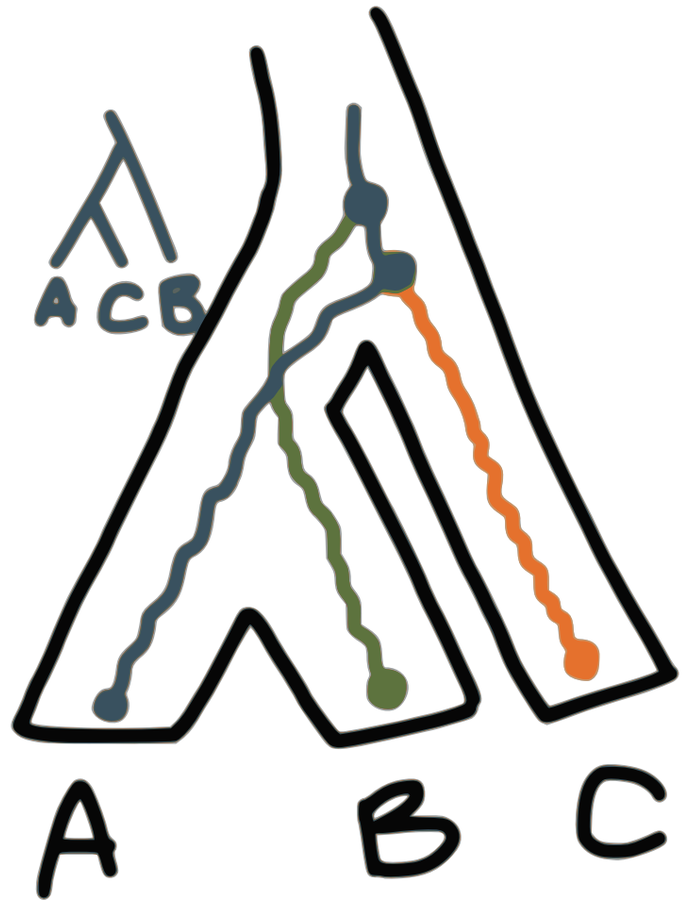
$$= 1 - \frac{2}{3} e^{-t}$$

Multispecies coalescent on a tree

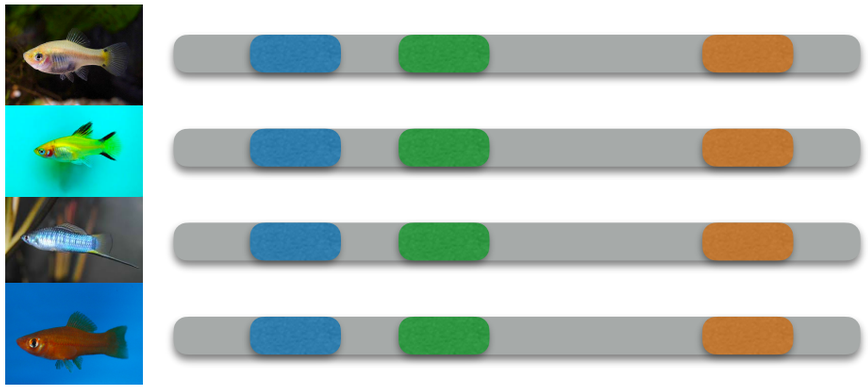


$$1 - \frac{2}{3}e^{-t}$$

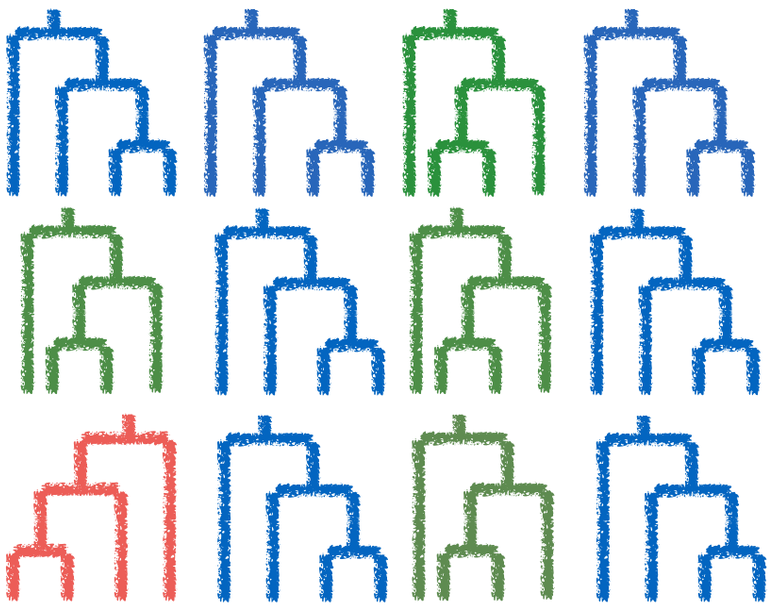
$$\frac{1}{3}e^{-t}$$



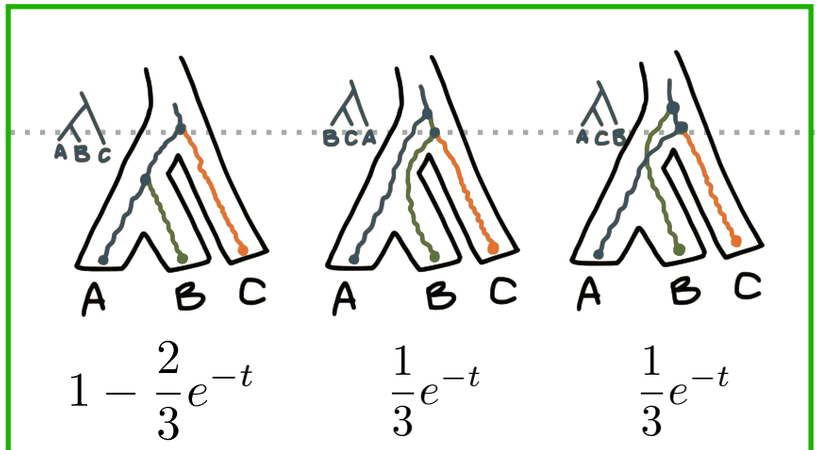
$$\frac{1}{3}e^{-t}$$



Distances
Parsimony
Likelihood
(Bayesian)

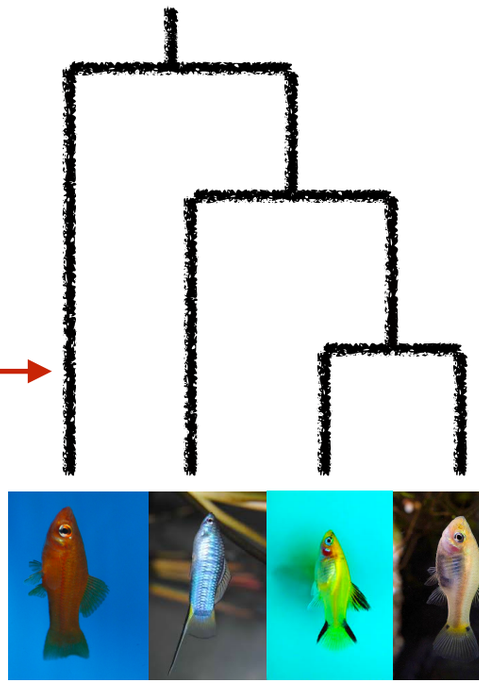


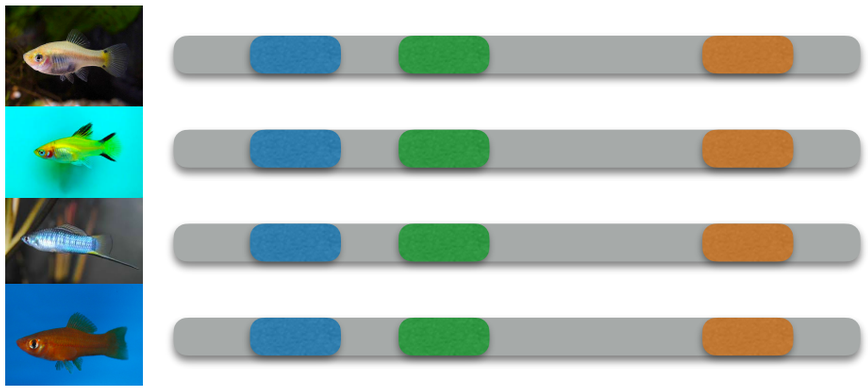
Data



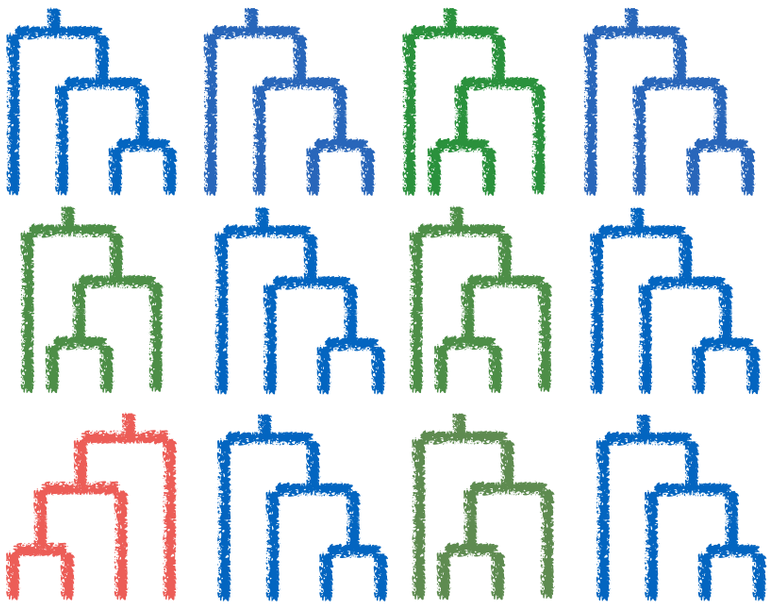
$$L(T,\theta) = \prod_{i=1}^{L} P(G_i|T,\theta)$$

Max. Lik.





Distances
Parsimony
Likelihood
(Bayesian)

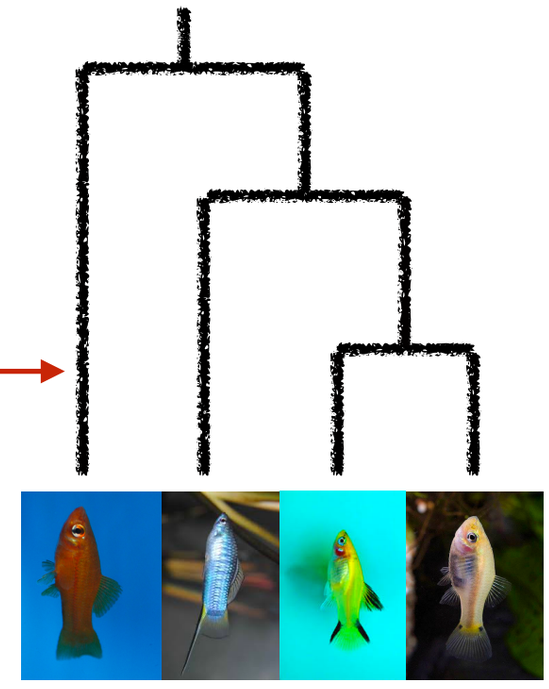


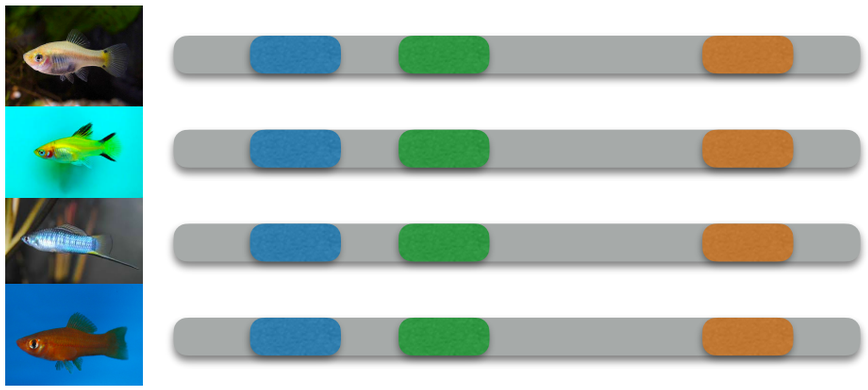
Data

$$L(T,\theta) = \prod_{i=1}^{L} P(G_i|T,\theta)$$

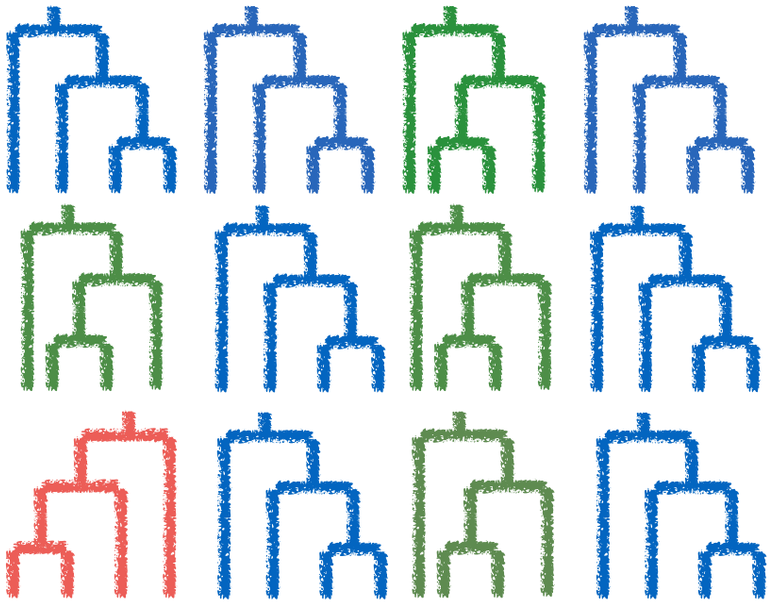
Max. Lik.

1. Guess a species tree
2. Evaluate likelihood of data (gene trees) given species tree
3. Search space of trees for species tree that maximizes likelihood





Distances
Parsimony
Likelihood
(Bayesian)



$$P(T,\theta|G) \propto \pi(T)\pi(\theta)\prod_{i=1}^{L} P(G_i|T,\theta)$$

Prior Tree

Multispecies Coalescent

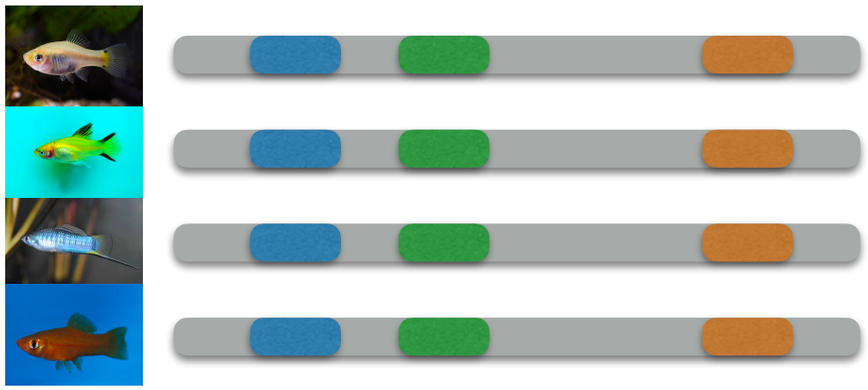
Bayesian

$$L(T,\theta) = \prod_{i=1}^{L} P(G_i|T,\theta)$$

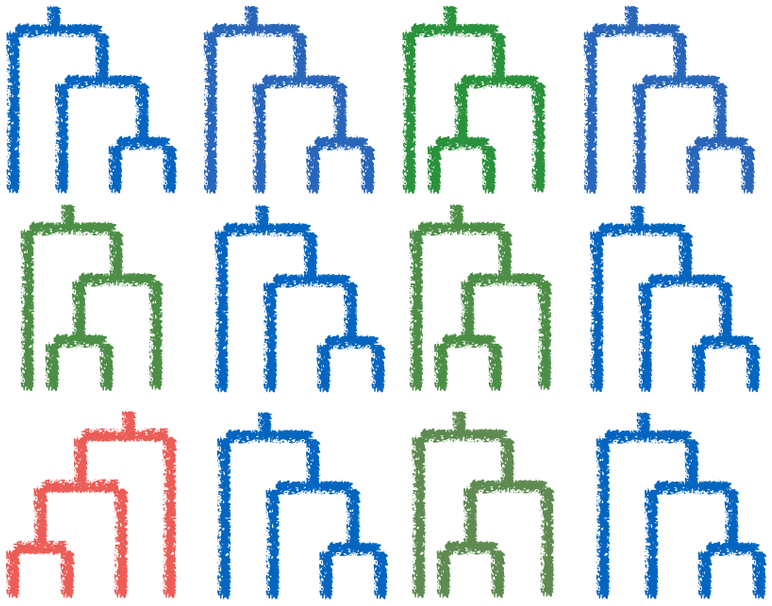
Max. Lik.

Summary methods: ASTRAL, BUCKy, MP-EST

(Zhang et al, 2018) (Larget et al, 2010) (Liu et al, 2010)



Distances
Parsimony
Likelihood
(Bayesian)



$$P(T,\theta|G) \propto \pi(T)\pi(\theta)\prod_{i=1}^{L} P(G_i|T,\theta)$$

Prior Tree

Multispecies Coalescent

Bayesian

$$L(T,\theta) = \prod_{i=1}^{L} P(G_i|T,\theta)$$

Max. Lik.

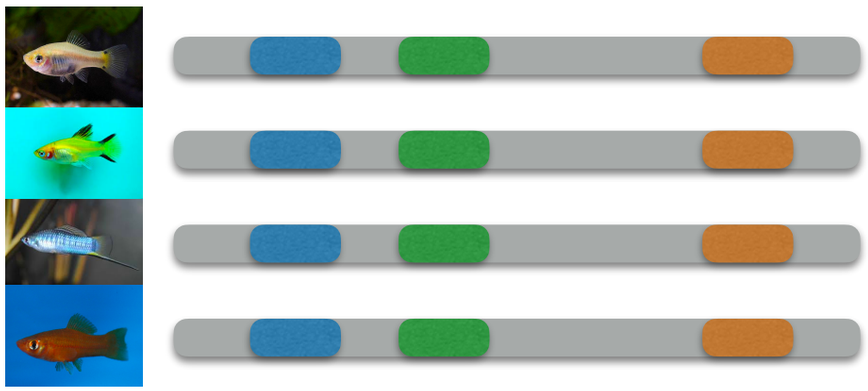
Summary methods: ASTRAL, BUCKy, (Zhang et al, 2018) (Larget et al, 2010)

Do not search tree space

Approx lik

MP-EST (Liu et al, 2010)

Co-estimation (lecture 15)



Species tree

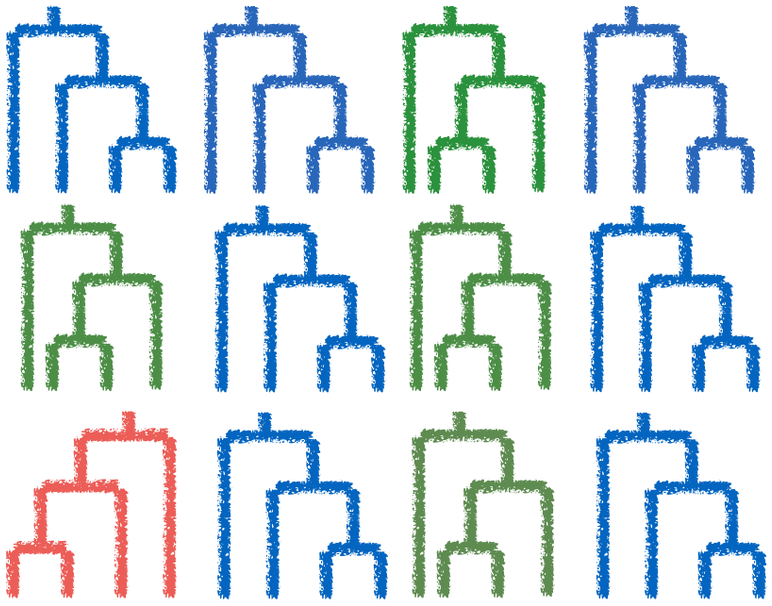
$$P(T, G, \theta | D) \propto \pi(T)\pi(\theta) \prod_{i=1}^{L} P(D_i|G_i) P(G_i|T)$$

gene trees; L loci

Prior Tree

Substitution Model

Multispecies Coalescent



$$P(T, \theta | G) \propto \pi(T)\pi(\theta) \prod_{i=1}^{L} P(G_i|T, \theta)$$

Bayesian

$$L(T, \theta) = \prod_{i=1}^{L} P(G_i|T, \theta)$$

Max. Lik.

Summary methods: ASTRAL, BUCKy, MP-EST

(Larget et al, 2010) (Liu et al, 2010)

Anomaly zone

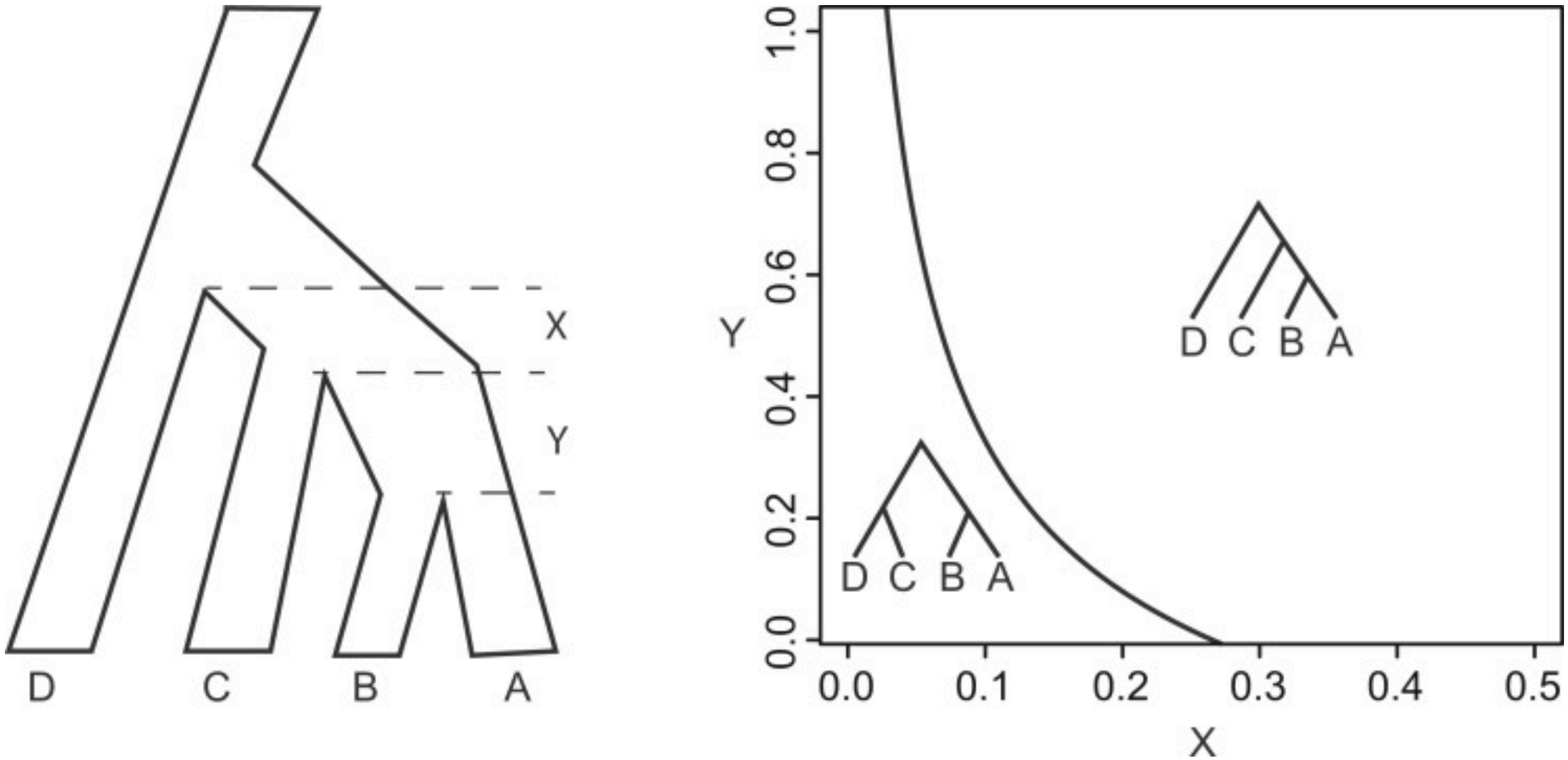
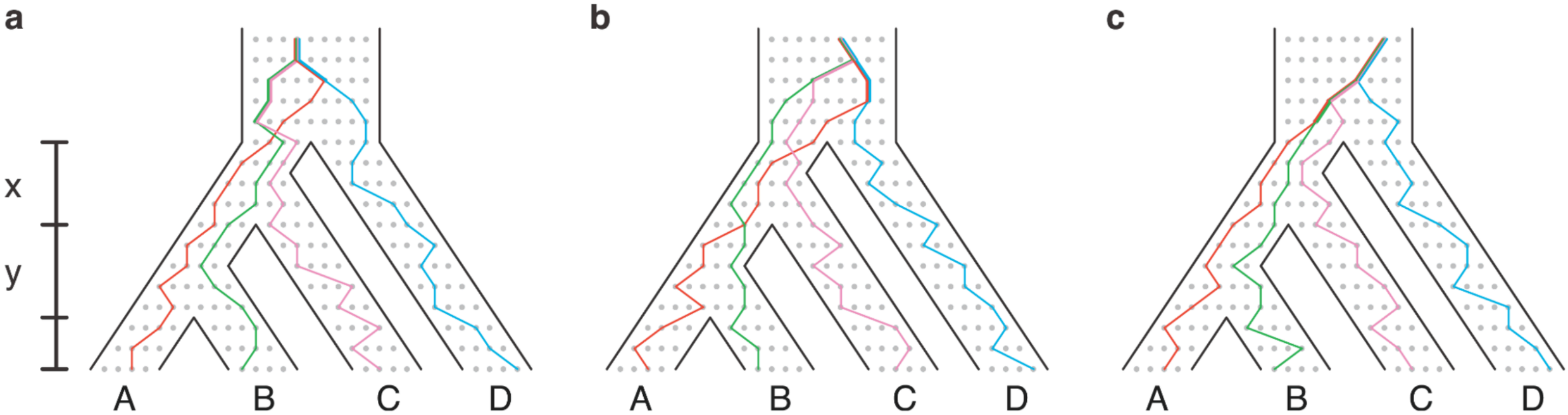
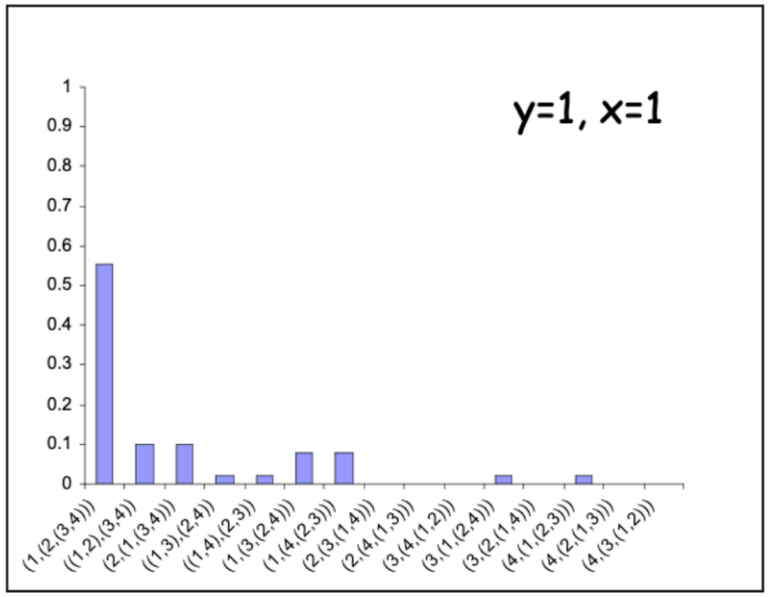


Figure 1 in Linkem et al (2016)



If the internal branches of the species tree—x and y—are short so that coalescences occur deep in the tree, the two sequences of coalescences that produce a given symmetric gene tree topology together have higher probability than the single sequence that produces the topology that matches the species tree. (a) and (b) Two coalescence sequences leading to gene tree topology ((AD)(BC)). In (a), the lineages from B and C coalesce more recently than those from A and D, and in (b), the reverse is true. (c) The single sequence of coalescences leading to gene tree topology (((AB)C)D). (Degnan and Rosenberg, 2006)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0.9
0.8
0.7
0.6
0.5
0.4
0.3
0.2
0.1
0
(1,(2,(3,4)))
((1,2),(3,4))
(2,(1,(3,4)))
((1,3),(2,4))
((1,4),(2,3))
(1,(3,(2,4)))
(1,(4,(2,3)))
(2,(3,(1,4)))
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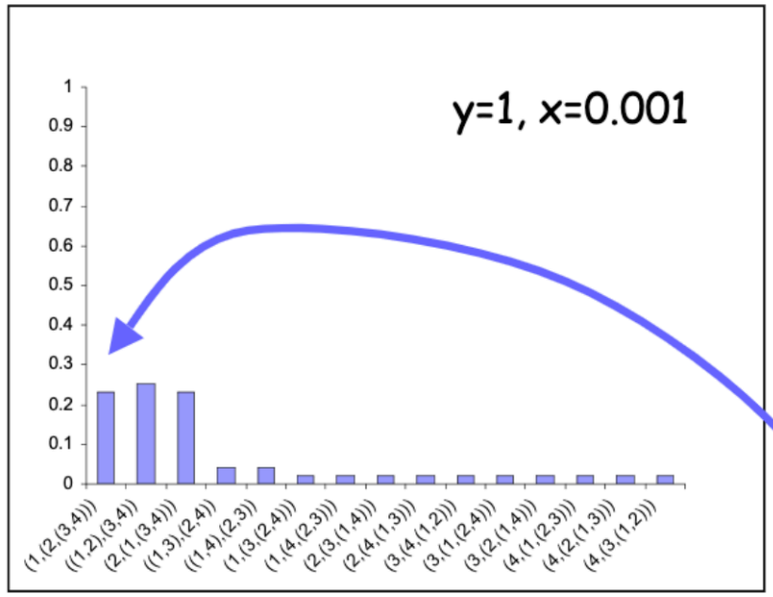
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0.7
0.6
0.5
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0.2
0.1
0
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((1,2),(3,4))
(2,(1,(3,4)))
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(1,(3,(2,4)))
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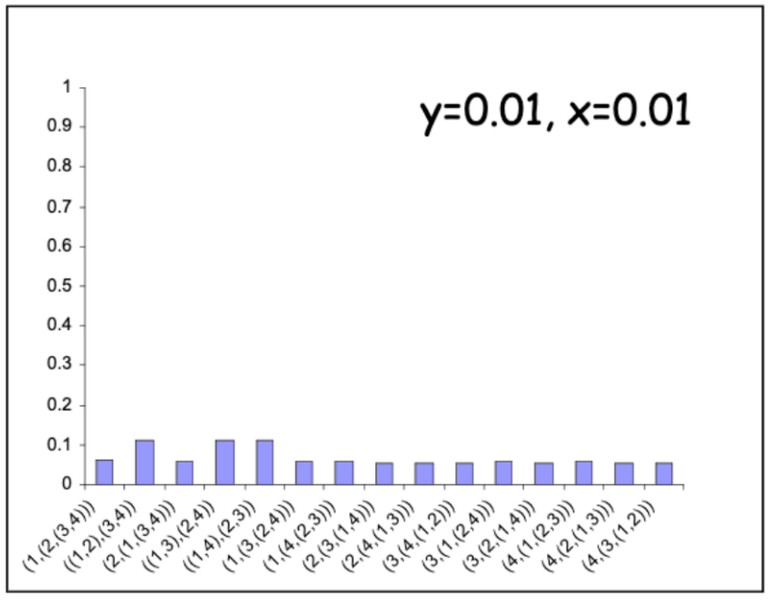
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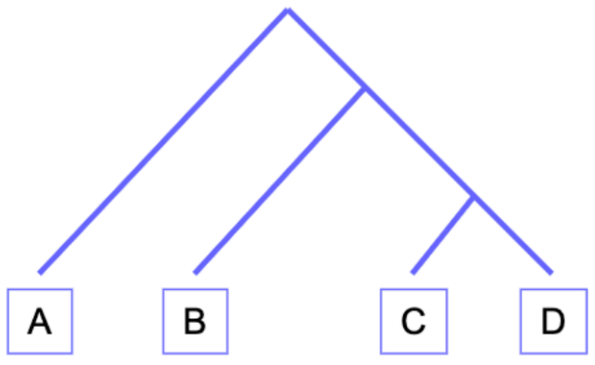
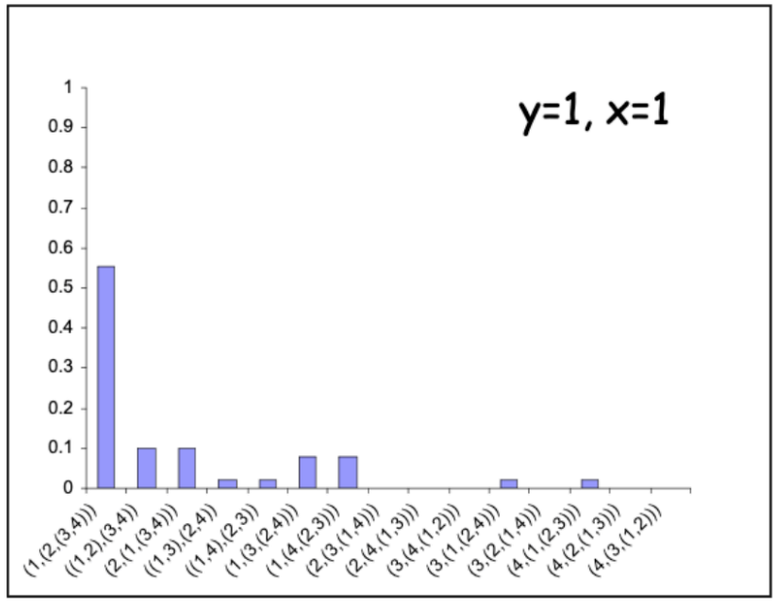

A
B
C
D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((1,3),(2,4))
((1,4),(2,3))
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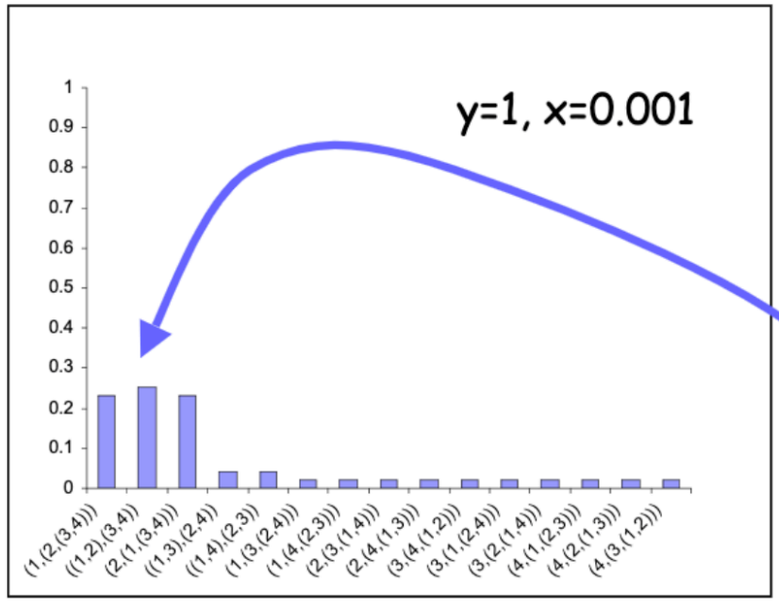
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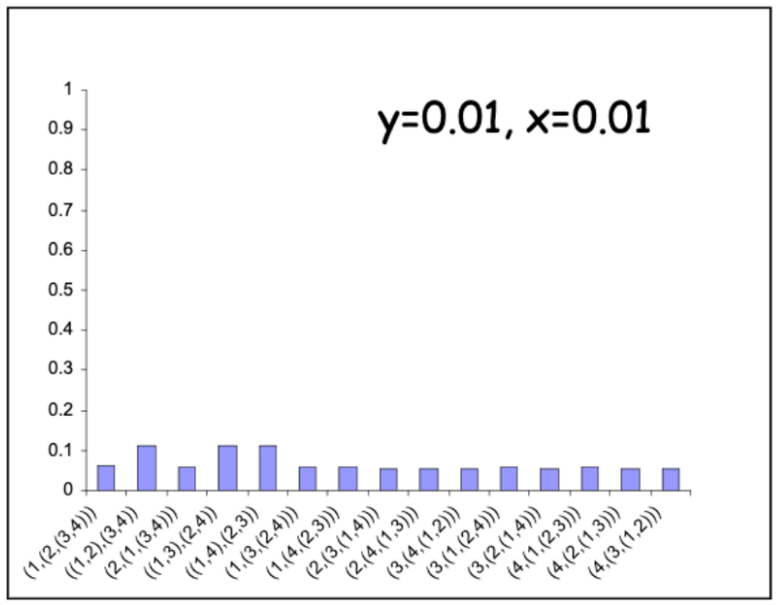
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0.1
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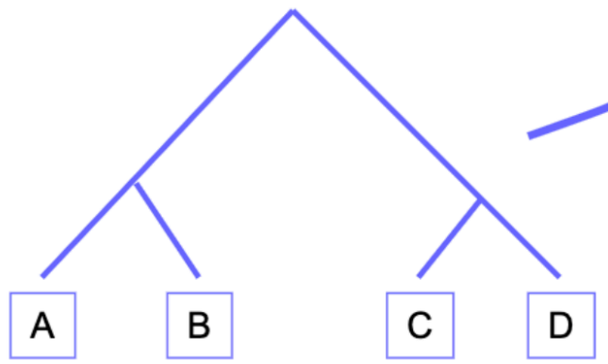
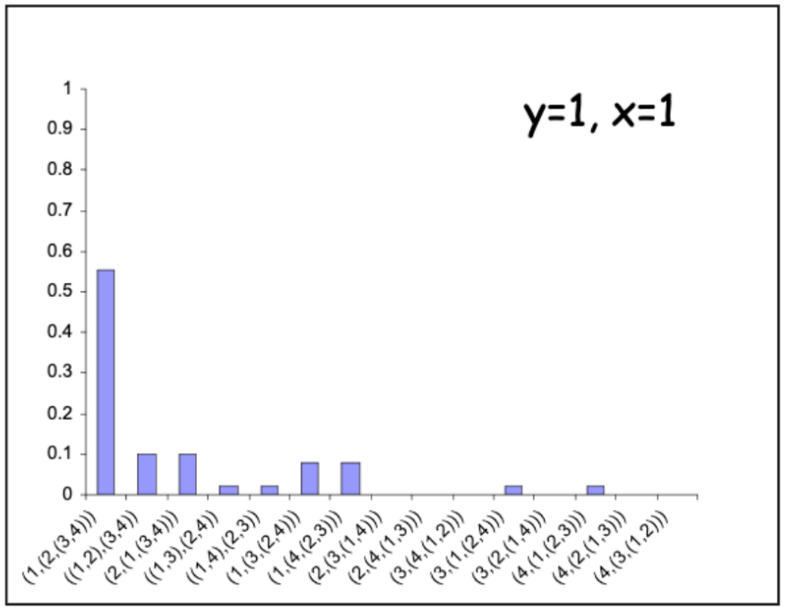

A
B
C
D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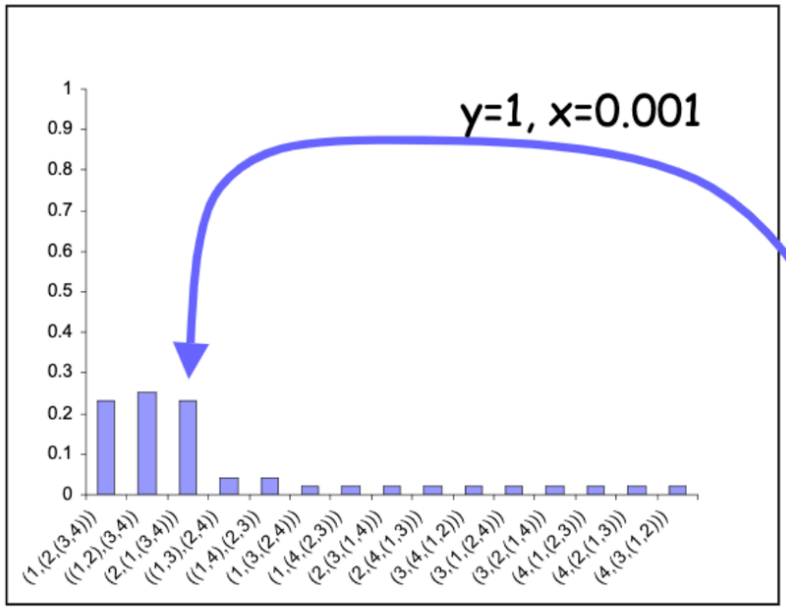
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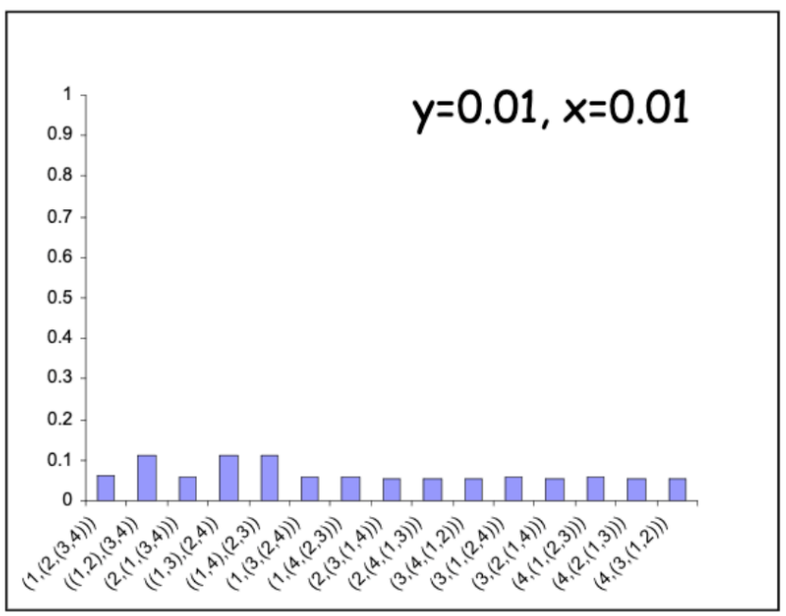
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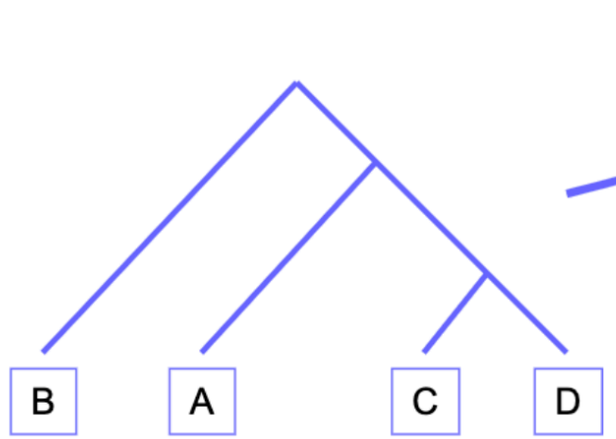
B
A
C
D

- Concatenation: assumes all genes follow the same tree-like history

- Coalescent-based tree methods: accounts for ILS (and sometimes gene tree estimation error), but does not allow for other sources of gene tree discordance (like GDL or gene flow)

- Extensions to coalescent-based tree methods:

    - Coalescent-based network methods: accounts for ILS, gene tree estimation error and gene flow

    - Coalescent+GDL methods (Li et al, 2020)

For next class:

- We will go over ASTRAL and BUCKy
- Each student is assigned to one software and has to read the paper for that software
- We will have a class discussion followed by installing and using the software